Face Embryology

Human eyes are highly sensitive in discerning minimal differences in the structure of human faces and in distinguishing persons. Discerning eyes can also detect many facial anomalies associated with systemic congenital anomalies. A classification of these facial anomalies based on embryological bases is helpful for understanding the morphogenesis of each anomaly.

In the normally developing embryo, the moment the human embryo is fertilized to the week of the baby's birth is an important period for human appearance. During preimplantation stages, differentiation occurs between precursors of embryonic and extraembryonic structures. A fore-hind axis begins within the inner cell mass at the time of implantation. By the end of the eighth week of gestation, the appearance of the head, face, hands, and feet suggest the embryo's species but is not yet definitive.

When nonhuman mammalian development is compared with human development, the study subjects must be compared at the same developmental stage (fetal, perinatal, postnatal) When collected appropriately, data from experimental studies of nonhuman mammalian embryos elucidate important aspects of human facial development.

Facial morphogenesis. Craniofacial development is an extraordinarily complex process that requires the orchestrated integration of multiple specialized tissues, such as the surface ectoderm, neural crest, mesoderm, and pharyngeal endoderm, in order to generate the central and peripheral nervous systems, axial skeleton, musculature, and connective tissues of the head and face. Understanding the development of the structures of the face also requires knowledge of the pharyngeal or branchial arches. These arches form on either side of the foregut and correspond to the primitive branchial arches. The pharyngeal arch consists of a core of mesenchyme covered externally by ectoderm and covered internally by endoderm.

The ectoderm is well around the stomodeum by the fourth week of embryonic development and contributes to the formation of the face and the nasal and oral cavities.

The mesenchyme that fills the pharyngeal arches is derived from the following 3 origins: the paraxial mesoderm, the lateral plate mesoderm, and the neural crest cells. Although paraxial mesoderm and lateral plate mesoderm contribute to the musculature that develops in each particular arch, neural crest cells contribute to the skeletal portion of each arch.

At the early stages of embryonic development, the vertebrate face has a common plan. A series of small buds of tissue called the facial primordia forms around

the stomodeum, which forms the primitive mouth. The facial primordia are made up mainly of neural crest cells that have migrated from the cranial crest and settled.

The upper jaw develops from the following 5 main buds of tissue: a single median frontonasal mass (sometimes present as the median nasal processes or frontonasal prominences), the 2 lateral nasal prominences on both sides, and, flanking these, the 2 maxillae (maxillary prominences). The lower jaw develops from the paired mandibular primordia (mandibular prominences). Paired maxillary and mandibular prominences are derivatives of the first pair of branchial, or pharyngeal, arches. All of these prominences are produced by the proliferation of the neural crest cells that migrate into the arches from the neural crest during the fourth week of gestation.

The neural crest cells give rise to the connective tissue components, including cartilage, bone, and ligaments in the facial and oral regions. The myogenic cells of the muscles constitute a separate cell lineage. These cells originate from the paraxial mesoderm and migrate into the facial primordia. Prior to emigration, the neural crest cells in the head are formed according to which facial primordium they belong.

The individual facial primordia are populated by neural crest cell populations that arise in different regions of the head neural folds. The neural crest cells that settle to form the frontonasal mass first migrate from the prosencephalic region (forebrain) and are later joined by other migrating cells, mainly from the anterior mesencephalic region (midbrain). The cells of the maxillae come from the posterior mesencephalic region, whereas the cells of the mandibular primordia come mainly from the region of the anterior rhombencephalon (hindbrain). Cells that arise in the posterior mesencephalon also contribute. In the trunk, exchanges between different regions of the neural crest almost invariably lead to normal development.

The frontonasal prominence surrounds the ventrolateral part of the forebrain, which gives rise to the optic vesicles. These vesicles project from the sides of the forebrain into the mesenchyme and form the eyes. The frontal portion of the frontonasal prominence forms the forehead, whereas the nasal part of the frontonasal prominence forms the rostral boundary of the stomodeum and nose.

A summary of the derivatives of the prominences is as follows:

- Frontonasal prominence - Forehead and the dorsum apex of the nose
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- Lateral nasal prominences - Sides (alae) of the nose
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- Medial nasal prominences - Nasal septum

- Maxillary prominences - Upper cheek region and most of the upper lip
- Mandibular prominences - Chin, lower lip, and lower cheek regions
- Mesenchyme in the facial prominences - Fleshy derivatives and various bones

A summary of the derivatives of the first and second pharyngeal (ie, branchial) arches is as follows:

- Pharyngeal arch I
    - Cranial nerve - Maxillary and mandibular division of the trigeminal nerve (cranial nerve V)
    - Artery - Maxillary (terminal branch)
    - Muscles - Muscles of mastication (ie, temporalis, masseter, pterygoids), mylohyoid, anterior belly of digastric, tensor tympani, and tensor veli palatini
    - Skeleton - Maxillary cartilage (incus, alisphenoid), mandibular or Meckel cartilage (malleus), and arch dermal mesenchyme (maxilla, zygomatic, squamous portion of temporal bone, mandible)
- Pharyngeal arch II (hyoid)
    - Facial nerve - Cranial nerve VII
    - Artery - Stapedial
    - Muscles - Muscles of facial expression (ie, orbicularis oculi, orbicularis oris, risorius, buccinator, platysma, auricularis, frontalis), stapedius muscle, posterior belly of digastric, and stylohyoid muscle
    - Skeleton - Stapes, styloid process, stylohyoid ligament, lesser cornu of hyoid, and the upper part of the body of the hyoid bone

Early development of the face. Facial development occurs mainly between the fourth and eighth weeks of gestation.

- Fourth week of development (stage 12 and 13)
    - Primordia of the face appear at the cephalic end of the embryo.
    - Two nasal placodes cap the bulbous frontal prominence.
    - The optic discs appear posterolateral to the frontal prominence.
    - Three paired branchial arches have formed.
    - The first arches split into maxillary and mandibular prominences. The hyoid arches are the second pair.
    - Between the first arches and frontal prominence, the buccopharyngeal membrane becomes fenestrated.
- Fifth week of development (stage 14 and 15)
    - Nasal pits develop in the nasal placodes, and the rims of the placodes differentiate into medial and lateral nasal prominences.
    - The lens vesicles invaginate and close within the optic discs.
    - The mesenchyme of the mandibular arch fills in across the midline.
    - The caudal end of the medial nasal prominences begins to fuse with the maxillary prominences.
- At the beginning of the sixth week of development (stage 16)
    - The nasals have shifted to a more ventral, central position.
    - Growing and shifting subectodermal mesenchyme smooths out the furrows between prominences and arches, and the second arch becomes more massive.
    - Six auricular hillocks, which will become the pinna of the ears, form on the mandibular and hyoid arches.
- By the end of the sixth week of development (stage 17)
    - Medial and lateral nasal prominences fuse.
    - Maxillary prominences begin the formation of the upper jaw.

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    - The midline approximation of the medial nasal prominences forms the nasal septum.
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- At the beginning of the seventh week of development (stage 18)
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    - The tip of the nose is elevated between the medial nasal prominences and is visible in profile.
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    - Eyelids become prominent.
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    - The pinna of the ear takes shape.
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- End of the seventh week of development (stage 18)
    -
    - The pattern of facial features has taken on a human appearance. However, facial proportions develop during the fetal period.
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    - The fusion of the medial nasal prominences, which forms the central axis of the nose and the philtrum of the lip, is complete.

Final development of the face. From the beginning of the eighth week of development to birth, the final facial development occurs slowly and consists mainly of changes in the proportion and relative positions of the facial components.

During the early fetal period, the nose is flat and the mandible is underdeveloped. They obtain their characteristic form while facial development is being completed. As the brain enlarges, it creates a prominent forehead, the eyes move medially, and the external ears rise.

The prenatal face is small because of (1) the rudimentary upper and lower jaws, (2) the unerupted primary teeth, and (3) the small size of the nasal cavities and maxillary sinuses.

Conclusion. The development of the vertebrate face is a dynamic multistep process that starts with the formation of neural crest cells in the developing brain and their subsequent migration to form, together with mesodermal cells, the facial primordia. Patterning and morphogenesis of neural crest–derived tissues within a developing vertebrate embryo rely on a complex balance between signals acquired by neural crest cells in the neuroepithelium during their formation and signals from the tissues that the neural crest cells contact during their migration. Neural crest cells carry information that directs the axial pattern and species-specific morphology of the head and face. Signaling interactions

coordinate the outgrowth of the facial primordia from buds of undifferentiated mesenchyme into the intricate series of bones and cartilage structures that, together with muscle and other tissues, form the adult face.

Some of the molecules thought to be involved have been identified through the use of mouse mutants, data from human craniofacial syndromes, and expression studies of signaling molecules during facial development. However, the way in which these molecules control the epithelial-mesenchymal interactions, which mediate facial outgrowth and morphogenesis, is unclear.

The role of neural crest cells in these processes has yet to be well defined. Similarly, the complex interaction of all these processes during face development and the candidate signaling molecules and their possible target genes have not been clearly defined.

Congenital Malformations, Neck

Introduction. Congenital masses in the neck include branchial cleft cysts, thyroglossal duct cysts (TGDCs), ectopic thymus cysts, dermoid and teratoid cysts, cystic vascular abnormalities, and lymphatic malformations such as the cystic lymphangioma.

Most neck masses in children are benign inflammatory lesions, which can be successfully treated medically with antibiotics. Most neck masses in children that require surgery for diagnostic and therapeutic purposes are congenital in origin. TGDCs and branchial cleft cysts are the 2 most common congenital lesions.

Baer first described the branchial apparatus in 1827. The branchial arches begin to develop during the second week of gestation. During the fourth week of fetal development, 5 ridges, known as branchial arches, form on the ventrolateral surface of the embryonic head. Each arch contains mesoderm from which cartilage, muscle, and bone develop. Each arch is separated from the other arches by an external cleft of ectodermal origin. Each arch also has an internal pouch of endodermal origin and an associated cranial nerve, artery, and cartilage.

The anatomic location of a branchial cleft abnormality represents the presumed branchial cleft of origin. Branchial cleft anomalies can manifest in several different forms, including the following:

- Fistula - Epithelial-lined tract with both an internal and an external opening
- Sinus - Incomplete fistula with either an internal or an external opening
- Cyst - Epithelial-lined cavity with neither an internal nor an external opening

More than 90% of branchial cleft anomalies arise from the second branchial cleft system. Approximately 8% of branchial cleft anomalies arise from the first branchial cleft system. Cysts arising from the third and fourth branchial cleft system rarely occur.

Branchial cleft anomalies and TGDCs occur with equal frequency in males and females. Branchial cysts are twice as common as either branchial sinuses or fistulas. TGDCs are the most common mass found in the midline of the neck in children.

Embryology. Branchial apparatus. During the third week of development, the flat trilaminar embryo undergoes a series of complex folds that result in the formation of a cylindrical embryo. During this time, the laterally placed clefts, known as branchial clefts, appear. These clefts are due to flexion folds of the fetus within the amniotic cavity. The basic tissues of development within the head and neck (ectoderm, endoderm, mesoderm, neuroepithelium) become organized into the pharyngeal apparatus, also known as the branchial apparatus, which is the forerunner of the head and neck structures.

Development of the branchial apparatus begins during the second week of gestation and is complete by week 6-7. The apparatus consists of 5 mesodermal (branchial) arches appearing in the lateral wall of the foregut and separated from each other externally by ectoderm-lined brachial clefts and internally by endoderm-lined pharyngeal pouches. The 5 arches, which are prominent in the lateral profile, are numbered from cranium to cauda as 1, 2, 3, 4, and 6. The fifth arch, which is buried, is called the sixth arch by convention.

The endoderm of the primitive pharynx transiently contacts the ectoderm to form thin, double-layered, branchial membranes at the caudal aspect of each cleft. The mesoderm then separates the ectodermal and endodermal layers of the membranes.

During the fifth week of development, the first arch and cranial portion of the second arch thicken and enlarge caudally. The caudal portion of the second arch, as well as the third and fourth arches, becomes submerged in the cervical sinus of His, a shallow ectodermal pit. The epipericardial ridge developing from the mesoderm lateral to the sixth arch enlarges the depth of the pit. From this tissue arise the infrahyoid muscles, the sternocleidomastoid-trapezius muscle complex, the muscles of the floor of the mouth, and the muscles of the tongue. Also contained in this ridge are the spinal divisions of the accessory nerve and the hypoglossal nerve. During further development, the sinus of His becomes obliterated and, in the adult, represents the angle between the dorsal surface of the strap muscles and the anterior margin of the sternocleidomastoid.

The human embryo has 5 pairs of pharyngeal pouches. The fifth of these is often considered an appendage of the fourth pouch. During development, the pouches

extend away from the pharyngeal wall and communicate with the pharynx via the elongated pharyngobranchial duct.

The primordia of the thymus arise from ventral saccules of the lining epithelium of the third pharyngeal pouch, with a small portion arising from the fourth pouch. During the sixth week of development, the saccules begin to elongate caudally, connecting to the pharynx via the thymopharyngeal ducts. As development continues, the connections of the thymic primordia migrate caudally and medially, and the saccules become obliterated by proliferating epithelium. By 8 weeks' gestation, the thymic primordia of each side unite, attach to the pericardium, and descend into the anterior mediastinum. At this point, the thymopharyngeal ducts degenerate, and the thymus attains its final position in the chest.

Aortic arches

Each branchial arch contains an associated aortic arch connecting the paired dorsal and ventral aortas. All 6 pairs of arches are not present at the same time, with the first 2 involuting prior to the appearance of the sixth.

The first arch gives rise to the maxillary artery. The dorsal segment of the second arch forms the stapedial artery. The third arch forms the common carotid artery, as well as the proximal portion of the internal carotid artery. The right fourth arch forms the proximal portion of the right subclavian artery, while the left fourth arch contributes to the aortic arch proper. The fifth arch arteries involute. The sixth pair forms the pulmonary arteries.

Thyroid gland

The thyroid gland begins developing during the first and second week of intrauterine life and is completed by week 11. It arises simultaneously from 3 bodies, the median anlage and 2 lateral anlagen.

The median anlage arises as endodermal thickening from the midline of the ventral pharyngeal wall, between the first and second branchial arches, at the junction of the developing anterior and posterior tongue. This is the site of the foramen cecum.

The median thyroid anlage comes in contact with the aortic sac of the heart. As the sac descends, it pulls the median thyroid caudally, causing the thyroid to accumulate more endodermal cells from the surrounding tissue. As the median thyroid anlage descends, its pharyngeal connection elongates as a stalk, namely, the thyroglossal duct, which normally disappears by the fifth to sixth week. At the end of 7 weeks, the median thyroid reaches its final position over the trachea and consists of a small isthmus and 2 lateral lobes.

The contribution of the lateral thyroid anlagen remains controversial. They are thought to receive contributions from the fourth and sixth branchial pouches. As the lateral anlagen migrate anteriorly, they detach from the pharynx and fuse with the median anlage, contributing up to one third of the volume of the gland.

The parafollicular cells, which produce calcitonin, are derived from neural crest cells. They become incorporated in the lateral thyroid primordia, forming the ultimobranchial bodies.

Lymphatic system

Several theories exist pertaining to the development of the lymphatic system.

The first theory, known as the venous origin with centrifugal spread theory, centers on the development of "sprouts" off of large central veins. These then enlarge, coalesce, and form new sprouts that eventually form the lymphatic system. The direction of spread is toward the periphery.

The second theory, known as the mesenchymal origin with centripetal spread theory, describes the independent development of the lymphatic system from the confluence of mesenchyme spaces. The spaces move centripetally by interacting with other mesenchymal spaces.

The final theory, known as the combined venous-mesenchymal origin theory, states that the lymphatic system originates from the confluence of small venules with spaces in the mesenchyme. In this theory, development progresses through distinct phases.

- The first phase begins at approximately 7 weeks of intrauterine life with the development of endothelial buds, known as the lymphatic primordia, sprouting from veins and uniting to form plexuses. These give rise to paired axillary and jugular sacs.
- The second phase displays enlargement of these sacs and the development of single communications with the ipsilateral internal jugular veins, contralateral internal jugular veins, and axillary sacs.
- The third stage shows enlargement and widening of these sacs and the appearance of other lymphatic primordia (eg, internal thoracic primordia, paratracheal primordia).
- Further enlargement continues in the fourth stage, with coalescence of the primordial sprouts.
- During the fifth and final stage, all lymphatic primordia fuse, and one continuous system is formed, with permeation of the lymphatic channels into the tissues. The formal thoracic duct and cisterna chyli take shape. Development is complete by 10 weeks.

Branchial malfomations. Congenital anomalies of the neck arise as a consequence of disturbances in the complex development of the branchial apparatus of the fetus. They are classified according to their branchial cleft or pouch of origin as well as their anatomic relationships. They may take the form of a fistula, sinus, or cyst, based on the degree of completion of development of the anomalous structure.

Fistulae represent persistence of both the cleft and the corresponding pouch, thereby forming a communication (ie, fistula) that is epithelial lined. The fistula lies caudal to the structures derived from that particular arch and connects the skin to the foregut. Sinuses may be considered partial fistula, usually opening externally, with no internal opening. They are epithelial lined. Fistulae and sinuses may be lined by stratified squamous, columnar, or ciliated epithelium, and they may contain lymphoid tissue as well.

Cysts have neither an internal nor an external opening and are most often lined by stratified squamous epithelium (ectoderm derived); however, they can be lined by columnar epithelium (endoderm derived from pouches).

Classification of branchial anomalies

First branchial anomalies. First branchial anomalies represent 1% of all branchial anomalies. Work reports the following 2 types:[1]

- A type I branchial anomaly is ectodermally derived and is a duplication of the external auditory canal (EAC). Type I branchial anomalies may manifest posteriorly, adjacent to the pinna and concha. They may terminate near a bony plate at the level of the mesotympanum.
- Type II branchial anomalies contain both ectoderm and mesoderm. They are more common and can originate along the EAC, middle ear cleft, or nasopharynx. Type II branchial anomalies often manifest with a fistula emanating from the concha, EAC, or neck, with a tract extending medially and inferiorly to the EAC. They may pass medial or lateral to the facial nerve and terminate at the level of the anterior border of the sternocleidomastoid muscle. Often an opening is found in the EAC as well as below the jaw.

First branchial anomalies do not usually involve the middle ear or tympanic membrane.

Patients with first branchial anomalies may present with unilateral facial paralysis. First branchial anomalies may be associated with hemifacial microsomia.

Second arch anomalies. Second arch anomalies are the most common and represent 90-95% of branchial anomalies.

They are classified into 4 categories (I-IV) based on anatomic location. Types I-III are the most frequently occurring second arch anomalies, with type II being the most common. Bilaterality of second arch anomalies is uncommon.

- Type I anomalies are located along the anterior margin of sternocleidomastoid muscle at the junction of the middle and lower thirds, deep to the platysma and cervical fascia.
- Type II anomalies lie in contact with the great vessels.
- Type III anomalies pass medially between the internal and external carotid arteries, extending toward the lateral pharyngeal wall and lying above the glossopharyngeal and hypoglossal nerves and below the stylohyoid ligament.
- Type IV anomalies are very rare and are located next to the pharyngeal wall, medial to the great vessels at the level of the tonsillar fossa.

Cystic lesions are more common than fistulae. They tend to manifest as smooth, soft masses in the lateral neck and are located anterior and deep to the sternocleidomastoid muscle. Fistulae tend to manifest as recurrent neck infections, often following an upper respiratory tract infection, below the level of the digastric muscle. The most common time for presentation of second branchial anomalies is during the second decade of life.

Branchiootorenal syndrome (BOR syndrome), or *Melnick-Fraser syndrome*, is an association of auricular malformations, branchial fistulae, deafness, and renal anomalies. An estimated 2% of profoundly deaf children have BOR syndrome. The estimated prevalence is 1 per 40,000 people. External ear malformations may include preauricular pits, tags, lop-ears, and/or microtia. Anomalies of the ossicles, facial nerve, and fallopian canal have also been described.

Third branchial anomalies. Third branchial anomalies are rarely encountered. Many authors agree that differentiating between third and fourth branchial anomalies on clinical grounds is difficult.

A complete fistula has a cutaneous opening along the anterior border of the sternocleidomastoid muscle. The tract courses posterior to the common and/or internal carotid artery, superior to the hypoglossal nerve, and inferior to the glossopharyngeal nerve; then it medially pierces the posterolateral aspect of the thyrohyoid membrane to open into the pyriform sinus.

Third branchial anomalies can manifest with upper airway compromise in the neonate and may also manifest with hypoglossal nerve palsy.

Fourth branchial anomalies. Fourth branchial anomalies are extremely rare and, unlike second branchial anomalies, typically manifest in childhood.

Fourth branchial anomalies originate at the apex of the pyriform sinus, traveling anteriorly and inferiorly to the cricothyroid muscle and thyroid cartilage. A lateral cervical cyst with an internal fistula in the pyriform sinus is a common occurrence. The course of the tract then runs superiorly over the hypoglossal nerve and descends in the neck posterior to the common carotid artery.

Fourth branchial anomalies follow a different course on each side of the neck. On the right side, the tract passes underneath the subclavian artery and courses superior to the recurrent laryngeal nerve and inferior to the superior laryngeal nerve. On the left side, the tract passes anteriorly underneath the aorta and courses superiorly in the neck, posterior to the common carotid artery.

Fourth branchial anomalies often terminate in the perithyroid space, thyroid gland, or cervical esophagus, and they may manifest clinically as an abscess in these areas. The cutaneous opening is along the anterior border of the sternocleidomastoid muscle.

Clinical manifestation. Branchial cleft cysts manifest in a different manner than branchial sinuses and branchial fistulae. The typical branchial cleft cyst, in the absence of infection, manifests as a nontender, smooth, round mass located along the anterior border of, or just deep to, the sternocleidomastoid muscle. Depending on the arch of derivation, the location can be anywhere from the external auditory canal to the clavicle. Branchial cleft cysts usually enlarge gradually and often are not detectable until the second or third decade of life.

Branchial cleft cysts that manifest in early childhood usually occur with an acute and painful enlargement of the cysts secondary to an upper respiratory infection. Branchial cleft cysts commonly increase in size in the presence of an upper respiratory tract infection and then decrease in size as the infection resolves.

An infected branchial cleft cyst can progress into an abscess or rupture spontaneously to form a draining sinus tract. Neonatal patients and patients with larger cysts can present with aerodigestive tract compromise and associated symptoms of stridor, dyspnea, and dysphagia. Second and third branchial cleft cysts can cause stridor with life-threatening airway obstruction in neonates.

In contrast to those patients with branchial cleft cysts, patients with branchial cleft sinuses and fistulas often present soon after birth because the external opening is visible on the skin. Branchial cleft fistulas and sinuses may also be palpable as a fibrous cord extending along the anterior border of the sternocleidomastoid muscle. Mucus drainage may occur from the external opening. Secondary infections from cutaneous organisms, most commonly *Staphylococcus aureus* and group A beta-hemolytic streptococci, cause purulent drainage and erythema and edema.

An infected first branchial cleft sinus or fistula may cause aural drainage in the absence of otitis media or otitis externa. A child presenting with both a draining ear and a tender mass in the neck may rarely have a first branchial cleft cyst associated with a sinus or fistula tract. A child presenting with a mass in the neck and a draining tract along the anterior border of the sternocleidomastoid muscle may have a second or third branchial cleft anomaly. A child with paratracheal swelling associated with tenderness and fever may have acute suppurative thyroiditis. Multiple authors have documented the spread of bacteria from an internal opening in the pyriform sinus in third branchial cleft anomalies.

Diagnosis and management of branchial anomalies. Diagnosis is based on physical examination, including a thorough otologic examination. This should include the use of a Lempert or Vienna bivalve ear speculum to examine the cartilaginous EAC. Furthermore, flexible laryngoscopy may reveal a sinus tract in the pyriform sinus. For patients with suspected pyriform sinus fistulae, perform a barium esophagram.

Radiologic confirmation is best achieved using computed tomography. Typically, the cysts are of low attenuation with smooth walls.

Aspiration of the cystic mass may aid in confirming the diagnosis, thus excluding a neoplastic or inflammatory lesion.

Management of branchial cleft anomalies is surgical excision. Surgery should ideally be performed on the uninfected neck. Institute a course of antibiotics prior to the surgical treatment if infection is suspected.

Special considerations.Each of the branchial cleft types has special considerations.

- For first branchial anomalies, the potential for facial nerve involvement must be considered. Exploration of the nerve is mandatory before excision of the tract.
- For second branchial anomalies, the tract usually bisects the external and internal carotid arteries just above the bifurcation, traveling laterally and superiorly to the hypoglossal and glossopharyngeal nerves and terminating in the tonsillar fossa. These are typically addressed via an incision along the anterior border of the sternocleidomastoid muscle, although an intraoral approach is possible for isolated pharyngeal cysts.
- For third branchial anomalies, the tract travels posteriorly to the internal carotid artery, superiorly to the hypoglossal nerve, and inferiorly to the glossopharyngeal nerve. External approaches along the sternocleidomastoid muscle are preferred.

- Approach fourth branchial anomalies through a traditional thyroid incision. Pay special attention to the course of the recurrent laryngeal nerve.

The most common complication of surgical resection is recurrence, which occurs in approximately 3% of cases.

Thyroglossal duct cysts. Thyroglossal duct cysts (TGDCs) represent the most common congenital anomaly of the neck, accounting for 2-4% of all neck masses. They are most commonly present in the first decade of life but may also be seen in adults.

TGDCs are caused by a persistent epithelial tract, the thyroglossal duct, during the descent of the thyroid from the foramen cecum to its final position in the anterior neck. They may also give rise to sinuses, fistulae, or cysts.

TGDCs manifest in 6 different varieties. The infrahyoid type accounts for 65% of TGDCs and is mostly found in the paramedian position, while the suprahyoid type accounts for nearly 20% and is positioned in the midline. Juxtahyoid cysts make up 15% of TGDCs. Intralingual location occurs in approximately 2% of TGDCs, and the suprasternal variety occurs in approximately 10% of cases. Intralaryngeal TGDC is very rare. This variety has recently been described in a 42-year-old man and should be differentiated from intralaryngeal lesions.

TGDCs are usually nontender and mobile. Infected TGDCs may manifest as tender masses with associated dysphagia, dysphonia, draining sinus, fever, or increasing neck mass. They often manifest after an upper respiratory tract infection. Airway obstruction is possible, especially with intralingual cysts. Ultrasound and CT scanning are the radiologic tools of choice. Ultrasound can distinguish between solid and cystic components. CT scanning may reveal a well-circumscribed cystic lesion, 2-4 cm in diameter with capsular enhancement. A fistulogram may reveal the course of the tract. Thyroid scanning is suggested to rule out the possibility of the cyst containing the only functioning thyroid tissue, albeit in an ectopic site.

The surgical treatment of choice for thyroglossal cysts is the Sistrunk operation, in which an en block resection of the sinus tract and above (including the midportion of the hyoid bone) is performed. Recurrence is approximately 3-5% and is increased by incomplete excision and a history of recurrent infections.

Ectopic thyroid. Ectopic thyroid tissue can arise from either the median anlage or lateral anlage (far less common). The tissue may or may not be functioning. Prevalence is higher in females than in males, with a female-to-male ratio of 7:1.

The most common site of ectopic tissue is within the tongue (90%). This is most often located in the midline dorsum, which is attributed to the descent of the thyroid through the foramen cecum, as discussed above.

The ectopic tissue is typically found incidentally, although these masses can be symptomatic and can cause dysphagia, dysphonia, stridor, dyspnea, hemorrhage, or hoarseness. An association has been noted between lingual thyroid tissue and cretinism.

CT scans often reveal a homogenous hyperdense mass within the lingual musculature. Management of the mass is surgical; however, prior to the decision to operate, perform thyroid scanning to assess if the mass is functioning and to determine if this tissue is the only viable thyroid material.

Hemangiomas. Hemangiomas represent the most common of all congenital anomalies, with an incidence of 0.3-2% at birth and 10% at age 1 year.

Hemangiomas result from the inappropriate development of vascular endothelium and channels and associated nervous components.

Classification of hemangiomas

- Type I - Forms of neonatal staining, including stork bite and nevus flammeus
- Type II - Intradermal capillary hemangiomas, including salmon patches, port wine stain, and spider angiomas
- Type III - Juvenile capillary hemangiomas, including strawberry marks, strawberry hemangiomas, and capillary cavernous hemangiomas
- Type IV - Arteriovenous fistulas
- Type V - Arteriovenous malformations are present. Most hemangiomas present within the first year of life and continue to grow until approximately age 2 years, after which involution occurs.

The new classification according to Mulliken differentiates between hemangiomas and vascular malformations. They differ primarily in that hemangiomas are proliferative lesions while vascular malformations are vessel malformations, which enlarge only by dilation of the involved vessels.

Hemangiomas are classified by location as either superficial (above the dermis), deep (below the dermis), or mixed. Although a large number of hemangiomas will involute, many (as high as 50%) may require treatment. Primary treatment is high-dose steroids during the proliferative phase (usually in the first year of life), followed by surgical management later. Surgical management may involve a combination of both laser treatments and surgical resection.

Vascular malformations are classified according to the type of blood vessels involved. In contrast to hemangiomas, they grow by distentions of the vessels and not by proliferation. Types of vascular malformations are as follows:

- Venular - Small venules (capillaries) in the dermis (formerly classified as capillary malformations)
    - The most common type is the port wine stain.
    - Midline venular lesions (also known as stork-bites) tend to involute by age 1 year.
- Venous - Incorrectly classified as cavernous hemangiomas
    - These consist of dilated veins.
    - Their growth later in life is due to dilation of the veins.
- Lymphatic - Also known as lymphangioma or cystic hygroma
    - These lesions form from dilated lymph vessels and may suddenly enlarge following infection or trauma.
    - Lymphatic malformations are subclassified into macrocystic lesions, microcystic lesions, and mixed lesions.
- Arteriovenous - These abnormal connections of arteries (high-pressure) and veins (low-pressure) progressively dilate because of high pressures transmitted to the venous side.

Mixed lesions, including different types of vessels (eg, venolymphatic), can also occur. Management of vascular malformations depends on the type of vessel involved. The most common sites of manifestation include the face, neck, and scalp.

Physical examination reveals a soft, painless, and compressible mass, which may be located within the skin or mucosa or within muscle, bone, or salivary gland tissues. A pale telangiectatic lesion often precedes hemangiomas, and an audible bruit may be present.

Some of the distinctive features include the following:

- Salmon patch - Light pink or rust colored, most common on the back of the neck, forehead, and eyelids
- Spider angiomas - Small central arteriole with radiating capillaries
- Strawberry mark - Pale halo, surrounded by telangiectasia
- Capillary hemangioma - Red or purple, well-defined borders, blanches with pressure
- Cavernous hemangioma - Red or purple, "bag of worms" texture, blanches with pressure, increases in size on Valsalva
- Port wine stain - Pink or red, purple in adulthood, do not blanch with pressure, underlying soft tissue hypertrophy

- Syndrome associated hemangiomas - Occur in Von Hippel disease, Maffucci syndrome, Sturge-Weber syndrome, and Kasabach-Merritt syndrome

The diagnosis of hemangiomas is made on clinical grounds. Radiologic confirmation can be achieved with Doppler ultrasound, which also determines the flow rate within the lesion. CT scanning is used to map the extent of the lesion. Angiography helps to distinguish between hemangiomas and vascular malformations.

Treatment of patients with these lesions can pose considerable difficulty in light of the potentially irreversible cosmetic defects. Spontaneous involution occurs in as many as 90% of cases, with the majority involuting by age 5 years. As such, observation is initially recommended.

Other treatment modalities could be used for the approximately 3-5% of patients with nonresolving lesions. These methods have different success rates. They include electrodesiccation, compression, embolization, cryotherapy, sclerotherapy, radiation, chemotherapy, and carbon dioxide and neodymium: yttrium-aluminum-garnet (ND: YAG) lasers.

Thymic anomalies. Thymic anomalies arise from the ventral saccules of the epithelium of the third pharyngeal pouch. Anomalies can result from (1) incomplete descent of the thymus into the chest, (2) sequestration of thymic tissue foci along the descent path, or (3) failure of the thymopharyngeal duct to involute.

This may result in an aberrant cervical thymus or thymic cysts. Sequestered cysts can occur along the path of the thymopharyngeal ducts from the level of the mandible to the chest. Occasionally, trapped thyroid or parathyroid remnants may be within the cysts.

Typically, these cysts are asymptomatic masses that are not found at birth but discovered later in life, with two thirds manifesting during the first decade of life. Males are affected more frequently than females are.

Patients most often present with an asymptomatic neck mass, which may be slowly increasing in size. The mass may grow rapidly, secondary to infection, hemorrhage, cystic degeneration, or venous engorgement. Approximately 10% of patients present with symptoms of mass effect and compression, including dysphagia, dyspnea, pain, or hoarseness. Thymic cysts can manifest in association with numerous other conditions, including thyrotoxicosis, aplastic anemia, Down syndrome, neurofibromatosis, and Hodgkin lymphoma, although this is quite rare. Patients may also present with myasthenia gravis, which is associated with neoplasms of the thymus. Transudative pleural effusion due to rupture of a thymic cyst into the pleural cavity is very rare. This was recently

documented in a patient who reported chest pain in the right hemithorax and dyspnea on exertion.

Sternocleidomastoid tumor of infancy. Sternocleidomastoid tumor of infancy (SCTI) is part of the spectrum of conditions known as congenital muscular torticollis (CMT). It represents the most common neck mass of the immediate perinatal period, within the first 2 months of life. The term tumor is a misnomer because most commonly it is congenital fibrosis within the muscle. The etiology remains unclear, with the most likely cause being peripartum injury. SCTI has a slight male prevalence and occurs more often on the right and with breech presentations. SCTIs are typically a firm, painless mass with fusiform shape, approximately 1-3 cm in length. They are most often located in the inferior to the middle third of the sternocleidomastoid muscle, affecting the sternal or clavicular heads equally.

Diagnosis is based on CT scanning and ultrasound confirmation. Open biopsy is reserved for cases that do not resolve. The natural course of the disease is a peak in growth for approximately 8 weeks, with complete resolution by 6 months. Physiotherapy is recommended to achieve full range of motion. Some patients have small areas of residual fibrosis. Surgery is reserved for patients in whom torticollis is present for more than one year, those in whom craniofacial asymmetry develops, and those for whom physiotherapy fails. Long-term follow-up care is essential because of the possibility of recurrence or reappearance of the disease during periods of heightened growth.

Cervical teratomas. Cervical teratomas are extremely uncommon lesions of the head and neck. The prevalence of cervical teratomas is 1 in 16,000 individuals, most of them occurring in full-term, preterm, or stillborn infants.

In the neck, they most often occur on the anterolateral surface, extending midline from the thyroid gland. They may manifest clinically as severe respiratory distress and dysphagia due to compression in the proximity of the trachea and esophagus.

These lesions are often large, extending as far as 12 cm in their longest axis. Histologically, they may contain any combination of tissues from the 3 germ layers.

They are often categorized in relation to their proximity to the thyroid gland. The categories include (1) teratomas of the thyroid gland, deriving their blood supply from the thyroid arteries, (2) teratomas adjacent to the thyroid gland, which displace the gland but in which a definitive blood supply cannot be identified, and (3) teratomas of the neck. This categorization is of little clinical use. An extremely rare case of fetal cervical teratoma presenting at 24 weeks of gestation was recently published. In this case, a submaxillary mass and agenesis

of corpus callosum were revealed with ultrasonography and also associated with a subarachnoid cyst.

Cervical teratomas are managed with early surgical excision. This is particularly important in light of the possible malignant degeneration of these lesions. The prognosis in nonneoplastic cases is excellent and approaches 100%. The ex-utero intrapartum treatment (EXIT) procedure is a technique designed to allow partial fetal delivery via caesarean section with establishment of a safe fetal airway. The most common indication for the EXIT procedure is the presence of fetal airway obstruction, which is usually caused by a prenatal diagnosed neck mass.[2]

Dermoid cysts. Dermoid cysts are considered the most common form of teratoma and are characterized by a predominance of ectodermal content. Most dermoids in the head and neck occur in the region of the floor of mouth, with approximately one fourth of dermoids involving the lateral neck or midline regions.

They are believed to be caused by epithelial rests trapped during embryologic development.

Clinically, they manifest as slow-growing lesions that produce symptoms when their cystic lumens become filled with keratin debris. Sebaceous material may also be found in the cyst, alluding to its ectodermal origin.

The management of these cysts is surgical excision.

Midline cervical cleft. Midline cervical cleft (MCC) is a rare congenital anomaly. Fewer than 100 cases have been reported overall, the first being described by Bailey in 1924. It is not considered a true cleft because no skin gap exists

The etiology is unclear and is believed to be abnormal fusion of the second and third arches or nonfusion at the level of the ectoderm.

MCC typically manifests at birth with a cleft extending from the inferior aspect of the mentum to the level of the suprasternal notch. Often the area has serous drainage, and the superior aspect of the cleft takes on the appearance of a pseudonipple.

Physical examination and CT scanning are used to make the diagnosis. Distinguishing this entity from other midline neck masses (eg, thyroglossal duct cysts, bronchogenic cysts) is important. In general, midline cervical clefts are located caudally, while thyroglossal duct cysts are located cranially. Bronchogenic cysts are located in the suprasternal notch, with possible extension into the mediastinum.

Associated defects may be a median cleft of the mandible, tongue, and lower lip. A delay in mandibular development and hypoplasia or absence of neck structures, such as the hyoid bone may be associated. Associated thyroglossal and bronchogenic cysts may occur, as well as defects in other parts of the body such as a sternal cleft.[3] Management involves complete excision of the cleft with surgical repair of the soft tissue defect.

Lymphangiomas. Lymphangiomas are benign multiloculated cystic masses that are soft, compressive, and painless. They usually manifest at or shortly after birth, with 50-60% manifesting by age 1 year and 90% by age 2 years. They are uncommon lesions, accounting for fewer than 5% of all congenital neck masses. The term cystic hygroma is synonymous with lymphangioma.

Approximately 50-75% of all lymphangiomas occur in the neck, most commonly in the anterior and posterior triangles. They can occur in the submental triangle, with extension into the floor of mouth. These masses tend to manifest as slow growing, painless masses with a doughy consistency. They may acutely enlarge following infectious processes (eg, upper respiratory tract infections). As a result of acute enlargement or chronic growth, the hygromas may cause respiratory and swallowing difficulties.

Radiologic diagnosis is often made using ultrasound imaging. CT scanning often reveals a multicystic lesion with well-defined boundaries. Rim identification may be difficult in previously uninfected cysts because the lesions do not follow planes of embryologic fusion. MRI may offer better soft tissue delineation than CT scanning. Repeated infection or hemorrhage causes increased signal attenuation. Lymphangioma is classified into 4 histologic subtypes: cavernous lymphangioma, capillary lymphangioma, cystic hygroma, and hemangiolymphangioma.

The management of choice is surgical excision. Other techniques (eg, cryotherapy, sclerotherapy) have met with only marginal success. Even with surgical excision, recurrence rates for lymphangioma are high, reported at 6-50%.

Cleft Lip

Introduction

History of the Procedure

Chinese physicians were the first to describe the technique of repairing cleft lip. The early techniques involved simply excising the cleft margins and suturing the segments together. The evolution of surgical techniques during the mid-17th century resulted in the use of local flaps for cleft lip repair. These early descriptions of local flaps for the treatment of cleft lip form the foundation of surgical principles used today.

Tennison introduced the triangular flap technique of unilateral cleft lip repair, which preserved the Cupid's bow in 1952. The geometry of the triangular flap was described by Randall, who popularized this method of lip repair. Millard described the technique of rotating the medial segment and advancing the lateral flap; thus, preserving the Cupid's bow with the philtrum. This technique has resulted in improved outcomes in cleft lip repair.

Problem

Cleft lip is among the most common of congenital deformities. The condition is due to insufficient mesenchymal migration during primary palate formation in the fourth through seventh week of intrauterine life. This results in disfigurement and distortion of the upper lip and nose. Cleft lip may be associated with syndromes that include anomalies involving multiple organs. Patients may have impaired facial growth, dental anomalies, and speech disorders (if a cleft palate is present), and they may experience late psychosocial difficulties.

Frequency

The incidence of cleft lip in the white population is approximately 1 in 1000 live births. The incidence in the Asian population is twice as great, whereas that in the black population is less than half as great. Male children are affected more often than female children. Isolated unilateral clefts occur twice as frequently on the left side as on the right and are 9 times more common than bilateral clefts. Combined cleft lip and palate is the most common presentation (50%), followed by isolated cleft palate (30%), and isolated cleft lip or cleft lip and alveolus (20%). Fewer than 10% of clefts are bilateral.

For parents with cleft lip and palate or for a child with cleft lip and palate, the risk of having a subsequent affected child is 4%. The risk increases to 9% with 2 previously affected children. In general, the risk to subsequent siblings increases with the severity of the cleft.

Etiology

Little evidence exists that links isolated clefts to exposure to any single teratogenic agent. The exception is the anticonvulsant drug, phenytoin. The use of phenytoin during pregnancy is associated with a 10-fold increase in the incidence of cleft lip. The incidence of cleft lip in infants born to mothers who smoke during pregnancy is twice that of those born to nonsmoking mothers. Syndromic clefts are those associated with malformations in other developmental regions, with reported frequencies ranging from 5-14%.

The most commonly recognized syndrome associated with clefts of the lip and palate is Van der Woude syndrome. This syndrome is an autosomal dominant disorder characterized by clefts of the lip and/or palate and blind sinuses, or pits,

of the lower lip. Clefts of the secondary palate alone are far more likely to be associated with syndromes than are clefts involving the lip alone or the lip and palate. Most cases of lip clefts are nonsyndromic and believed to be either multifactorial in origin or the result of changes at a major single-gene locus.

Pathophysiology

Development of the upper lip is characterized by fusion of the maxillary prominences with the lateral and medial nasal prominences. This process starts during the fourth week of gestation and is completed by the seventh week. Failure of mesenchymal migration to unite one or both of the maxillary prominences with the medial nasal prominences results in a unilateral or bilateral cleft of the lip, respectively.

Classification

No universally accepted classification scheme exists for clefts of the lip and palate. Veau categorized clefts into 4 classes, as follows:

1. Clefts of the soft plate alone
2. Clefts of the soft and hard palate
3. Complete unilateral clefts of the lip and palate
4. Complete bilateral clefts of the lip and palate

This classification scheme does not provide a means of classifying clefts of the lip alone and ignores incomplete clefts. Kernohan stripped–Y classification allows the description of the lip, the alveolus, and the palate. In this classification, the incisive foramen defines the boundary between clefts of the primary palate (lip and premaxilla) and those of the secondary palate.

Presentation

Clefts of the lip may manifest as microform, incomplete, or complete clefts. Microform clefts are characterized by a vertical groove and vermilion notching with varying degrees of lip shortening. Unilateral incomplete lips manifest varying degrees of lip disruption associated with an intact nasal sill or Simonart band (a band of fibrous tissue from the edge of the red lip to the nostril floor). Complete clefts of the lip are characterized by disruption of the lip, alveolus, and nasal sill.

Bilateral clefts are almost always associated with cleft palate, with 86% of patients with such clefts of the lip presenting with palatal clefts. Unilateral clefts of the lip are associated with palatal clefts in 68% of cases. Nasal regurgitation during suckling may indicate an associated cleft of the palate. All infants with clefts of the lip should have a complete head and neck examination, including careful examination of the palate as far as the tip of the uvula. The presence of a

bifid uvula, a translucent central zone in the velum, and a detectable notch of the posterior border of the hard palate indicate submucosal palatal cleft.

All patients with clefts are best referred to multidisciplinary cleft lip and palate centers. Persistent otitis media and middle ear effusions are associated with palatal clefts and warrant regular follow-up care. Depending on the preference of the surgical centers, the otolaryngologist may elect to perform myringotomy before or after definitive cleft lip and palate repair.

Most cases of lip clefts are nonsyndromic. Parents should be reassured and advised sensitively. At the initial visit, review feeding techniques carefully. For the infant, breastfeeding and the capacity to suck are difficult. However, breastfeeding may be possible with isolated clefts of the lip and the alveolus. For infants with palatal clefts, a variety of special bottles and nipples are available. Crosscut soft nipples made for premature infants facilitate feeding of infants with cleft palate. At the conclusion of the initial consultation, the parents and the infant should be comfortable with the feeding method.

Indications

Clefts of the lip are usually repaired in early infancy. Reassure and advise the parents that operative intervention is best carried out at age 2-3 months. The rule of 10 serves as a safe guideline, ie, body weight should be approximately 10 lb, the hemoglobin concentration 10 g/dL, and age greater than 10 weeks.

Relevant Anatomy

The typical unilateral complete cleft lip deformity results from both a deficiency and a displacement of the soft tissues, the underlying bony structures, and cartilaginous structures. An imbalance of the normal muscular forces acting upon the maxilla results in an outward rotation of the premaxillary-bearing medial segment and posterolateral displacement of the smaller lateral segment.

The inferior edge of the anterior nasal septum is displaced out of the vomerine groove into the noncleft nostril, and the anterior septum leans laterally over the cleft. The overlying columella invariably is short on the cleft side and distorted by the displaced caudal septum. In the nasal tip, the alar cartilage is characteristically deformed, and the medial crus is displaced posteriorly. The dome is separated from that of the noncleft side, and the lateral crus is flattened and stretched across the cleft. The axis of the nostril on the cleft side is characteristically oriented in the horizontal plane. This position is in contrast to the normal vertical axis of the nostril on the opposite side.

The muscular fibers of the orbicularis oris do not decussate transversely as in the normal lip; rather, they course obliquely upward, paralleling the cleft margin toward the alar base on the lateral side of the cleft and toward the base of the

columella medially. The philtrum on the cleft side is short, and the presumptive Cupid's bow peak is displaced superiorly. The vermilion is deficient on the cleft side of the medial element.

Complete bilateral clefts of the lip result from failure of the premaxillary segment to fuse with the lateral maxillary segments. Subsequent forward growth of the premaxilla, attached only to the vomer above, leads to its projection beyond the lateral segments. Within the isolated prolabium, the skin is foreshortened vertically, the white roll is underdeveloped, and the vermilion is deficient. The prolabium lacks muscle fibers, and the philtral ridges, the central philtral dimple, and Cupid's bow are absent. The bilateral cleft nasal deformity is characterized by flaring of the alar bases and wide separation of the domal segments of the alar cartilages. The columella is markedly shortened, causing the nasal tip to be depressed.

Contraindications

Coexisting medical conditions that would result in cardiopulmonary complications, bleeding disorders, infection, and/or malnutrition are all contraindications to surgery.

Treatment

Surgical Therapy

Depending on the width of the cleft, some centers may perform presurgical orthopedic management to reduce the width of the bony cleft and to align the maxillary arch prior to definitive lip repair. This may be accomplished by using external traction devices or passive orthodontic plates. Surgical lip adhesion may be used for wide clefts as an alternative to presurgical orthopedics. In lip adhesion, the soft tissues of the superior lip are united, essentially converting a wide complete cleft to an incomplete cleft.

Because each cleft is unique, definitive repair of the cleft lip should be individualized. Mirault was among the first to describe the technique of increasing lip length by using a small flap taken from the cleft side. However, this technique did not reconstruct the Cupid's bow. Le Mesurier later described the inset of a rectangular flap from the cleft side into a releasing incision on the noncleft side to create an artificial Cupid's bow. Tennison described the triangular flap to preserve the Cupid's bow. Randall later worked out the geometry of this flap, adding precision and reproducibility to the triangular flap technique. The technique creates an unnatural scar across the philtral column and flattens the philtral dimple. In addition, the triangular flap method does not address the nasal deformity.

Millard introduced the rotation-advancement technique in 1955, which overcomes many of the pitfalls of earlier techniques. The rotation-advancement

technique is the most commonly used method today for the repair of unilateral clefts. The technique preserves the Cupid's bow and the philtral dimple and improves nasal tip symmetry. The rotation-advancement lengthens the lip by means of a rotation incision that releases the medial lip element, allowing the Cupid's bow to rotate downwards into normal position. A small backcut may be used to further increase the length, if needed. The lateral lip element is advanced into the gap created by rotation of the medial element, thus completing reconstruction of the upper lip.

Preoperative Details

- Administer general endotracheal anesthesia.
- Monitor pulse, respiration, blood pressure (BP), ECG, and temperature.
- Gently hyperextend the neck to provide optimal exposure.
- Ensure appropriate padding of pressure points.
- Ensure corneal protection (eg, lubricant, taping of the eyelids).
- Place the patient on a warming blanket.

Intraoperative Details

Unilateral cleft repair

Skin markings

Center of Cupid's bow

1. Cupid's bow peak at the vermilion-cutaneous junction, noncleft side of the medial element
2. Proposed Cupid's bow peak, cleft side of medial element
3. Midline of columella
4. Alar base, noncleft side
5. Alar base, cleft side
6. Proposed Cupid's bow peak, lateral element
7. Tip of advancement flap
8. End of backcut

Key points

- The position of point 3 may be determined by transposing the distance between 1 and 2, such that the distance between 1 and 2 is equal to the distance between 1 and 3.
- The distance between the alar base and Cupid's bow peak on the noncleft side should equal that on the cleft side, ie, the distance from 2-5 is equal to the distance from 7-6.
- The difference between the distance from the columellar base to points 2 and 3 represents the deficiency in vertical length that must be gained to

level the Cupid's bow. Although the rotation incision allows point 3 to drop inferiorly, some vertical deficiency of the cleft side may remain. The added length may be gained by making a small backcut medial to the philtral column on the noncleft side. The advancement flap derived from the lateral element fills the opening created by the rotation incision and any backcut in the medial element; hence, the distance from 3-5 plus the added length gained by the backcut equals the distance from 6-7. Introduction of a small triangular flap from the lateral element into a small transverse incision in the lower part of the lip may also serve to lengthen the cleft side of the medial element and to improve the contour of the lip. The base width of this flap is equal to the height of the vermilion-cutaneous roll.

- The rotation incision curves gently from point 3 to the columellar base, hugging the columellar-lip junction, and stops medial to the philtral column on the noncleft side. Crossing the normal philtral column results in an undesirable elongation of the lip on the noncleft side. In the infant with a rectangular philtrum, this incision may be modified as described by Mohler.
- Place point 7 on the lateral element at a point level with the Cupid's bow peak on the noncleft side (point 2) and where the white roll remains well developed. Placing this point too far laterally produces an unnatural shortening of the lateral lip element, which results in a noticeable imbalance. Placing this point too far medially, where the white roll is poorly developed, results in a noticeable irregularity of the white roll. To gain some extra vertical height, if needed, point 7 may be moved 1 mm laterally and point 3 moved 1 mm medially.
- The advancement incision curves from point 7 to point 8, then a variable distance to point 9, depending on the amount of rotation needed to correct the flare of the displaced alar base.
- When possible, line up the point at the junction of the wet and dry vermilion; this point is also called the red line.

Repair of the orbicularis oris

Reorientation and repair of the orbicularis oris muscle bundles are essential for normal lip function and eversion of the lip border. Failure to adequately address the muscle at the time of lip repair results in abnormal motion or contour when pursing the lips and in a characteristic bulge in the lateral lip element. A variety of techniques for reorienting the orbicularis oris muscle fibers in unilateral clefts have been described, although the optimal method of muscle repair remains to be determined. Park advocates careful identification and precise reapproximation of the superficial and deep components of the muscle.

Primary nasal correction

In every case, reconstruct both the lip and the nose at the primary operation. Repair of the lip in infancy while delaying nasal repair until later in childhood is no longer appropriate. Reconstruction of the cleft nasal deformity remains the most challenging aspect of cleft surgery. Principles of primary nasal correction include the following:

- Wide undermining of the nasal skin on the cleft side, freeing the skin from the underlying nasal skeleton
- Elevation of the slumped alar cartilage on the cleft side to the normal level using internal or external suspension sutures
- Medial advancement of the lateral crus and alar base on the cleft side

Bilateral cleft lip repair

Prior to surgical repair, the use of presurgical orthopedic appliances can reduce significant premaxillary protrusion. The premaxillary segment varies considerably in size and in the extent of its protrusion. In incomplete clefts, attachment of the premaxilla to one or both lateral maxillary segments limits premaxillary protrusion. In complete clefts, retroposition of the premaxilla prior to definitive lip repair often is necessary. This procedure may be accomplished through presurgical orthopedics, using external traction devices or passive orthodontic plates. Retroposition of the premaxilla may also be accomplished through surgical lip adhesion. Surgical setback of the premaxilla, a technique popular in the 19th century, is associated with subsequent midfacial growth impairment and should be avoided.

Modifications of the Millard straight-line, banked, forked-flap technique currently are the most widely used methods for repair of bilateral cleft lip. These techniques work well for the repair of complete bilateral clefts and may be modified for the repair of incomplete and/or asymmetrical clefts.

Key points

- The prolabium is always used in reconstruction of the philtrum, even if severely deficient.
- The prolabial vermilion nearly always is deficient, and the prolabial white roll usually is indistinct. Therefore, in most cases, the prolabial mucosa is turned down to line the buccal alveolar sulcus; the tubercle and white roll are reconstructed using paired white roll–vermilion–orbicularis marginalis flaps from the lateral lip elements brought beneath the prolabium.
- The prolabium must not be left too wide and should rarely exceed 5-6 mm in width.
- The orbicularis peripheralis muscle is freed from its abnormal attachments at the alar bases and from the overlying dermis. This allows the muscle to be mobilized and reconstructed over the premaxilla.

- Anatomic positioning of the alar cartilages is performed at the time of primary lip repair.

Postoperative Details

- After surgery, feeding is resumed using a soft crosscut nipple.
- Infants remain hospitalized for intravenous hydration until oral intake is sufficient (usually 24 h).
- The suture lines are kept clean by gentle application of a dilute hydrogen peroxide solution, and a small amount of antibiotic ointment is applied to the repair 3 times daily and after feeding.
- If nonresorbing suture material is used, the sutures are removed by the fifth postoperative day.
- Soft elbow restraints are used for 2-3 weeks to keep the infant from manually disrupting the repair.

Follow-up

Arrangements for suture removal are made prior to or immediately after discharge. All patients require long-term follow-up care. A dedicated multidisciplinary team approach and evaluation in different stages of the patient's life is important. Assess speech, language, hearing, somatic growth, and development regularly. Appropriately assess general dental health. Orthodontic management and secondary surgical procedures, such as bone grafting, are carried out during the school years. Patients with significant midface retrusion may require treatment. Secondary procedures to correct the tip in nasal asymmetry may be performed at school age; however, if the reconstruction involves osteotomy, delay the procedure until the completion of nasal growth (age 16-17 y). Emotional difficulties may emerge because of poor self-esteem during adolescence and should be recognized and addressed early.

Complications

Complications following cleft lip repair are unusual. Wound infections following surgery are uncommon and are treated with appropriate antibiotic therapy. Although immediate wound dehiscence is best repaired prior to discharge, treat delayed dehiscence after the scar has settled.

Cleft Palate

Author: Michael J Biavati, MD, Clinical Assistant Professor, Department of Otolaryngology, University of Texas Southwestern
Coauthor(s): Gina Rocha-Worley, CCC/SLP, MS, Speech Pathologist, Department of Speech Pathology, Harlem Hospital Center
Contributor Information and Disclosures

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Introduction

Cleft palate is a congenital deformity that causes a multitude of problems and represents a special challenge to the medical community. Special care is needed for patients with cleft palate. Speech production, feeding, maxillofacial growth, and dentition are just a few important developmental stages that may be affected.

Multiple specialists make up the team that works together to improve the quality of life for patients with cleft palate. Otolaryngologists, oromaxillofacial surgeons, plastic surgeons, nutritionists, and speech pathologists are just a few of the members of the team. Psychological effects on both the patient and the parents are important aspects that also need to be addressed. A significant number of patients with cleft palate have associated syndromes that may result in cardiac, limb, or other system defects. Although cleft palate deformity was described hundreds of years ago, to this day, no agreed-upon management algorithm exists for patients with cleft palate.

History of the Procedure

The first recorded operation on a palate was performed in 500 AD for inflammation of the uvula. For centuries, literature and interest in clefts were lacking because the deformity was thought to be due to syphilis.

Misconception also existed regarding the genetic predisposition. In 1887, the following passage was published in *Lancet:*

George Williamson stated that "[the fourth] law of heredity that [imposes] hideous physical impressions on the mind of a mother are [sic] capable of producing deformity and monstrosity in the offspring." As evidence, he introduced a report by Dr Child that described "a mother attending a penny show, where a trained horse pulled the trigger of a pistol, pretending to shoot a rabbit. A dummy was thrown out; back of its head was bleeding, having all the appearance been shot off." The woman bore a child resembling a rabbit.

Pare first described the use of obturators for palatal perforations in 1564. In 1552, Jacques Houllier proposed that the cleft edges be sutured together. However, it was not until 1764 that LeMonnier, a French dentist, performed the first successful repair of a cleft velum. Dieffenbach closed both the hard palate and the soft palate in 1834. von Langenbeck first described cleft palate closures with the use of mucoperiosteal flaps in 1861. In 1868, Billroth thought that fracturing the hamulus would enable better outcomes in surgery. Further modifications of the von Langenbeck technique came from Gillies, Fry, Kilner, Wardill, Veau, and Dorrance. The debate over the timing of closure led to a short break in early surgical repair. However, in 1944, Schweckendiek again began closing cleft defects in young patients.

Frequency

Cleft deformities of the palate are among the most common congenital malformations. A cleft palate can be diagnosed as early as the 17th week of gestation by means of ultrasonography. Although many studies exist, the exact environmental and genetic factors that play a role are still largely unknown.

The overall incidence of cleft palate with or without cleft lip is 1 case in 1000 live births. The incidence of cleft palate varies by race, with the highest rate among American Indians, at 3.6 cases per 1000 live births, and the lowest rate among African Americans, with 0.3 cases per 1000 live births. Among the total number of clefts, 20% are an isolated cleft lip (18% unilateral, 2% bilateral), 50% are a cleft lip and palate (38% unilateral, 12% bilateral), and 30% are a cleft palate alone. The incidence of isolated cleft palate (without cleft lip) is 1 case in 2000 live births. Submucous cleft palate is more common, with an incidence of 1 case in 1200-2000 patients, depending on the study population. Bifid uvula occurs in 1 of 80 patients and often occurs in isolation, with no clefting of the palatal muscles.

No racial predilection exists for cleft palate, with an equal incidence among all races. Although cleft lip and palate together occur more commonly in males, isolated cleft palate is more common in females.

Etiology

Palate formation begins at the end of the fifth week of gestation. At this stage, the palate consists of 2 parts, namely, the anterior (primary) palate and the posterior (secondary) palate. The medial nasal prominences form the intermaxillary (premaxillary) segment, which comprises the primary palate and incisor teeth. The primary palate extends posteriorly to the incisive foramen.

The secondary palate, which is formed by the lateral palatal processes, begins at the incisive foramen and contains a bony section and a muscular section. The lateral palatine processes appear at about the sixth week of gestation. They comprise the deep portions of the maxillary prominence that form 2 horizontal structures or palatal shelves, which ultimately are derivatives of the first branchial arch. These shelves are originally on either side of the tongue. As the tongue moves downward in the seventh week of gestation, the lateral processes grow medially. Fusion of the hard palate begins anteriorly and continues posteriorly in the eighth week of gestation.

A number of processes are involved in the fusion of the 2 processes. Programmed cell death at the free edges and production of a sticky coat of glycoproteins and desmosomes provide an ideal bonding surface interface. The left side tends to lag behind the right side, leading to a propensity for left-sided clefts. The nasal septum subsequently grows downward into the newly formed palate. The process is completed between the 9th and 12th weeks of gestation.

Bone begins to form in the anterior palate first and extends posteriorly. The soft palate and the uvula, which make up the posterior portion of the secondary palate, develop during the eighth week of gestation. The tensor veli palatini develop, followed by the musculus uvula. These structures are completed by the 17th week of gestation.

The genetic basis of cleft deformity is most likely heterogeneous and multifactorial. Autosomal recessive, autosomal dominant, and X-linked inheritance patterns have been described. For all parents, the odds of having a child with a cleft are 1 in 700. In families in which no first-degree relatives are affected, the recurrence rate for a cleft lip or palate in subsequent children is 2.5%. When one first-degree relative is affected, the rate of recurrence is 10%. Similar recurrence rates (10-12%) occur in offspring of persons born with cleft deformities. If the cleft is part of an autosomal dominant syndrome, the recurrence rate can be as high as 50%. A cleft deformity is associated with a syndrome in 30% of cases. More than 400 syndromes with a cleft deformity as one of the characteristics have been described.

As previously mentioned, the etiology of the cleft palate is not well understood; however, some evidence exists that external factors may play a role. Relatively few of the many recognized teratogens cause cleft palates. Alcohol consumption in the embryologic period does result in many infants with clefts. Other teratogens associated with cleft palates include phenytoin, retinoids, and illegal drugs (eg, cocaine). Mechanically induced clefts can occur in utero by means of direct impingement on the embryo.

Genetic mapping of families with inherited forms of cleft palate has resulted in the identification of genes involved in palate development. Cleft palate associated with ankyloglossia, an X-linked disorder, was shown to be caused by mutations of the *TBX22* gene. *TBX22* is a member of the T-box gene family, which are transcription factors in vertebrates involved with mesoderm direction. Specifically, *TBX22* is expressed in the palatal shelves just prior to their elevation above the tongue. Mutations in this gene result in cleft palate due to loss of *TBX22* function.

Presentation

Most overt clefts of the hard palate and/or the soft palate are discovered at birth and are often manifested by feeding difficulties. Suckling may be compromised by the loss of an oral seal on the nipple. Cleft palate, especially when associated with mandibular hypoplasia (as with a Pierre Robin sequence), may also cause airway difficulties because the tongue prolapses through the cleft into the nasal cavity and the posterior oropharynx.

Partial clefts of the soft palate or submucous clefts may be overlooked in neonates because they may be asymptomatic. Early manifestations include nasal

reflux of liquids or food. Later, as speech develops, hypernasal speech or nasal emission may result.

Indications

Major clefts of the hard palate and/or the soft palate are repaired surgically before the patient is aged 1 year. Instances in which this does not occur include those with complicating medical conditions, such as congenital heart disease or airway compromise. Cleft repair is deferred for cardiac conditions that may be compromised by a change in upper airway resistance. When upper airway obstruction is a major problem, such as with a Pierre Robin sequence, a tracheotomy may be necessary. Cleft repair can then be accomplished with a secure airway.

When a submucous cleft is present, the indications for surgery concern velar competence. Often, the decision to repair a submucous cleft palate is deferred until the patient is aged 4-5 years, when speech development is sufficient to determine the degree of hypernasality and the effect of the cleft on intelligibility. Cleft repair at this age may involve a pharyngeal flap, depending on the amount of velopharyngeal incompetence present.

Relevant Anatomy

The role of the palate is to provide a barrier between the nasal and oral portions of the respiratory tract. Velar actions with deglutition, respiration, and phonation are similar to those of a sphincter; hence, the velopharyngeal mechanism is often termed the velopharyngeal sphincter.

Familiarity with the anatomy of the palate is essential in understanding functional and surgical repair. Blood is supplied to the hard palate by the greater palatine artery, which enters via the greater palatine foramen. The lesser palatine artery and nerves pass through the lesser palatine foramen. Nerve supply originates from the maxillary branches from the trigeminal nerve, which forms a plexus that innervates the palatal muscles. Contributions from cranial nerves VII and IX enter posterior to the plexus.

The palatine aponeurosis is the principal structural element within the velopharynx. It provides an anchoring point for muscles, adding a degree of stiffness, and is continuous laterally around to the hamulus with the tensor veli palatini muscle. The aponeurosis is diamond shaped. More posterolaterally, the salpingopalatine ligament, the fascia of Tröltsch, and the internal fascia of the pharynx (which all form the membranous portion of the eustachian tube) contribute to the velopharynx.

The normal structure and function of the soft palate is dependent on the levator sling. This structure comprises portions of the tensor veli palatini, palatoglossal,

palatopharyngeal, and uvular muscles. Functionally, the levator veli palatini, palatoglossus, and musculus uvulae muscles either elevate the soft palate or alter its shape. Other muscles, such as the superior constrictor, palatopharyngeus, palatothyroideus, and salpingopharyngeus muscles, are involved with movements of the lateral and posterior pharyngeal walls. The tensor veli palatini is involved mainly with middle ear aeration. In patients with cleft palate, the muscle attachments are directed anteriorly and attach onto the posterior portion of the bony palate. These fibers must be surgically reoriented to achieve proper palatal function.

Treatment

Medical Therapy

A cleft palate is primarily a surgical problem, so no particular medical therapy exists for the condition. However, complications of a cleft palate, including feeding problems, airway obstruction, and otitis media, may require medical management prior to repair.

Feeding problems

Feeding a baby with a cleft lip and palate can be a challenge. Usually, a team of professionals is available to help the family meet this challenge by providing information regarding feeding and nutritional needs. A speech/language pathologist who specializes in feeding and swallowing disorders can provide the family with information regarding the most appropriate feeding position and equipment to use to make the feeding as normal as possible. An evaluation of the patient's swallowing skills for signs of aspiration or dysphagia is an important part of this assessment.

A nutritionist can help establish a feeding regimen that provides appropriate energy intake for optimal nutrition and growth. In general, a newborn needs 100-150 mL of breast milk or formula per kilogram of body weight per day. A prosthodontist or an orthodontist can construct an appliance to assist with feeding for infants who cannot tolerate prefabricated feeders. Since no separation exists between the oral and nasal cavities, children with a cleft palate (with or without a cleft lip) have difficulty obtaining adequate intraoral pressure for sucking and extracting liquid from the nipple. This can cause the baby to tire easily and to be unwilling or unable to suck long enough to obtain enough milk. In addition, food or liquid may back up and run out of the baby's nose and cause choking, coughing, or spitting up.

Families can make several modifications to help improve oral feeding. These modifications include using special cleft feeding equipment, properly positioning the infant during feeding, adjusting the placement of the nipple, supporting the cheek, and altering the rate of feeding.

Most babies with a cleft palate are fed with a bottle, although breastfeeding is not precluded and may be attempted in some cases. With bottle-feeding, the baby with a cleft palate typically feeds slower and needs help regulating the flow of liquid. Often, a chewing type of sucking is observed with use of the nipple. Various bottles and nipples can be used to assist with feeding.

A cleft feeding nipple must have a large enough opening to allow the formula to flow easily to prevent sucking fatigue, but it must not be so large as to cause choking. Nipples should be soft and compressible, allowing liquid to flow easily. Soft nipples designed for premature infants used with a regular bottle often work well. Occasionally, the hole in the nipple designed for a premature neonate may need to be enlarged to increase the flow of milk. This is best performed by creating an X-shaped opening to help regulate the flow of formula. Enlarging the opening too much may result in the free flow of milk, which can cause the baby to choke.

Several cleft palate feeders are available from several manufacturers. The Mead-Johnson Cleft Palate Nurser is the most commonly used bottle and nipple for babies with clefts. It includes a long soft nipple with a crosscut hole attached to a flexible plastic bottle that can be squeezed to increase the flow of liquid. The person feeding the infant can squeeze the bottle in conjunction with the baby's sucking/breathing rhythm to assist with swallowing. Squeezing timed with naturally occurring jaw compressions can also facilitate swallowing.

Other devices include the Ross Cleft Palate Nurser, manufactured by Ross Laboratories, that allows for a steady flow of liquid with minimal sucking. This nipple can be shortened to individualize the speed of the flow. The Haberman Feeder, manufactured by Medela, provides 3 flow rates determined by the position of the nipple in the oral cavity. The Haberman nipple is larger and longer than most nipples, and it can be gently squeezed to help the baby extract the liquid. To facilitate flow, a 1-way valve separates the nipple from the bottle. Air is squeezed out of the nipple before the feeding begins, and the valve allows the nipple to refill with liquid as it is squeezed or sucked. Reduction of air in the nipple helps decrease the baby's overall intake of air.

Placement of the nipple within the oral cavity is important. Optimally, the nipple should be positioned to the back and along the side of the mouth on the noncleft side (in the case of unilateral cleft palate). Supporting the cheek by gently squeezing the cheeks together around the nipple may also improve oral suction.

With regard to positioning, a semiupright position, as upright as possible, is best. This position helps prevent food and liquid from entering the nasal cavity. Upright positioning may also decrease eustachian tube reflux, which may lead to middle ear inflammation (otitis media). Eustachian tube reflux can also cause

otorrhea in children with ear grommets. An angle-necked bottle can make feeding in an upright position easier.

If a mother is interested, she should be encouraged to try breastfeeding. If the cleft affects only the lip and alveolar ridge and not the palate, breastfeeding may be successful. A baby with a cleft palate is not likely to obtain adequate suction to extract the milk. However, the difficulty varies and depends on how fast and easily the mother's milk flows; at the least, breastfeeding should be attempted to determine its success. The baby with a cleft lip and palate can be held with the cleft lip side next to the breast, as the soft breast may help create a lip seal not achievable with a regular nipple. With a good lip seal, the noncleft side may function more normally. Remember to position the baby as upright as possible.

Mothers who attempt breastfeeding must be aware of the signs of dehydration and the need to seek medical attention at the first sign of problems. Signs of dehydration in the baby include sleepiness and listlessness, urination fewer than 10 times per 24 hours, and urine that is strong smelling and/or dark and concentrated. In addition, the mother should carefully monitor the baby's weight and take into account the baby's frustration with feeding, sucking fatigue, and signs of hunger when deciding to continue or discontinue breastfeeding.

Frequent burping is important because babies with clefts tend to take in a lot of air with sucking. The gastroesophageal reflux may also be increased because of excessive air intake. Regular burping during feeding may help minimize spitting up. It may be beneficial to feed the baby smaller meals and to increase the number of feedings throughout the day, especially if the baby fatigues quickly with sucking. A nutritionist should carefully monitor these changes to ensure adequate energy intake for optimal growth. Ensuring that the mouth and lip area are clean after feeding and prior to placing the baby in a reclined position is important to prevent choking.

Spoon-feeding and feeding of textured and table foods usually evolves in the same quantities and developmental sequence as with any baby or child, even if the cleft palate remains open. Things to consider with spoon-feeding include presenting the food slowly, allowing the baby to remove food from the spoon by using its lips, and allowing the baby to regulate the timing of the next mouthful. For textured and table foods, the feeder should continue with the slow rate of presentation and provide sauce or gravy with small textured foods, such as rice, that tend to spread throughout the mouth. Adding sauce or gravy helps the pieces of food stick together as they are transported through the oral cavity.

Most babies and children with a cleft lip or palate learn to eat orally, with modifications. They become adept at moving the bolus through the oral cavity around the cleft. If food falls from the nose or gets stuck in the palate, the feeder should not become alarmed because the food does not interfere with breathing or

cause harm. Occasionally, the child may sneeze when food enters the nasal cavity. Food can be removed with a finger or a cotton swab without frightening the child. If the patient with cleft palate continues to have feeding difficulties even with the appropriate modifications, further consultation may be necessary to rule out such problems as dysphagia or sensory integration difficulties.

If the lack of weight gain due to feeding difficulties is a problem, use of a feeding tube should be considered. If problems with weight gain do not respond to feeding therapy, a gastrostomy may be necessary.

Airway obstruction

Airway obstruction may present in children with a cleft palate, especially those with mandibular hypoplasia (ie, a Pierre Robin sequence). Upper airway obstruction results from posterior positioning of the tongue, which is prone to prolapse into the pharynx with inspiration. Nasal obstruction can also result from the tongue protruding into the nasal cavity.

Airway obstruction is usually managed by placing the child in a prone position to prevent prolapse of the tongue. In severe cases in which the obstructed airway is not relieved with conservative measures, a tracheotomy may be necessary. In these instances, such measures as a lip-tongue adhesion are generally not as effective and not as well tolerated as a tracheotomy.

Otitis media

Otitis media is a common complication of a cleft palate and is present in nearly all children with unrepaired clefts. Although recurrent suppurative disease can be a problem, the primary complication is that of persistent middle ear effusion with resultant hearing loss. Medical management for this problem typically involves careful observation, which must be performed in light of the potential complications of prolonged hearing loss, especially in a child at risk for speech problems due to a cleft palate. In most instances, grommet insertion for middle ear ventilation is the preferred treatment to avert potential speech problems due to conductive hearing loss.

Surgical Therapy

General agreement exists that surgical correction of a cleft palate should be accomplished when patients are younger than 1 year, before significant speech development occurs. The potential benefits of an intact velum as a child begins to speak are believed to outweigh the possible complications of early closure, namely later collapse of the maxillary arch with a resultant crossbite.

How closure is accomplished is subject to some variation. Generally, 1-stage closure of the soft palate and/or the hard palate can be accomplished when the patient is aged 11-12 months. However, some advocate a 2-stage closure, with

repair of the velum (soft palate) when the patient is aged 3-4 months. This procedure results in narrowing of the hard palate cleft, facilitating closure at a later date, usually when the patient is aged 18 months. Similar to a lip adhesion for a wide cleft lip, a 2-stage approach may be useful when the cleft palate is particularly wide.

When cleft palate repair is deferred to later childhood or adulthood, repair often involves a pharyngeal flap. Incorporating a pharyngeal flap into the repair can help close a large defect and compensate for velopharyngeal dysfunction and speech problems.

The goal of repair in patients with cleft palate is to separate the oral and nasal cavities; this separation involves the formation of a valve that is both watertight and airtight. The valve is necessary for normal speech. The repair also helps with the preservation of facial growth and the development of proper dentition. Three factors that are considered necessary for satisfactory function of the soft palate for speech are adequate length, adequate mobility, and conformity of the dorsal surface to the pharyngeal wall. Most surgeons include levator muscle complex reconstitution as part of palate repair. Reconstruction of the muscle sling appears more important than anatomical retropositioning in terms of obtaining a dynamic functioning levator sling. However, not all surgical teams have accepted intravelar veloplasty.

If cleft lip is present, its repair can precede palatoplasty. Although early repair seems to have an advantage in decreasing the chances of speech delays, the risk for facial growth abnormalities and other midface-related problems may be increased. In the past, several criteria have been listed for patients undergoing any of the procedures. Some of these criteria include a hemoglobin level higher than 10 g/dL, weight gain, the absence of infection, and a full preoperative evaluation by a pediatrician.

Preoperative Details

Repair of the hard palate is not always possible when the soft palate is repaired, especially with wide bilateral clefts. The cleft size can decrease as much as 7% with growth in patients aged 3-17 months. The size can be further reduced with early repair of the soft palate (in patients aged 3-4 mo) followed by closure of the hard palate in patients aged nearly 18 months. This fact should be taken into consideration in planning the time and the type of the repair. The defect is usually smaller than it was originally when closure is performed after the soft palate defect has completely healed. The procedure can be performed in patients aged as young as 3 months, with a second procedure for closure of the defect when they are 6-12 months.

A great deal of debate exists regarding the timing of the repair. In the past, many surgeons believed that hard palate repair should be delayed until after eruption

of the molar teeth. Currently, most centers focus on completion of the cleft palate before the patient is 12 months. Debate had existed over whether or not delayed closure of the hard palate was beneficial or harmful to facial growth, but the evidence for either side has not been conclusive.

Intraoperative Details

Investigators in a multicenter study involving surveys of more than 300 surgical teams attempted to establish the common ground for repairs of cleft defects. Although no single technique was used universally, a trend has been established toward the use of earlier palate closure over the last several years. Of the closure techniques surveyed, the Furlow procedure was the most common technique for cleft palate closure. The basic surgical techniques included the following: von Langenbeck, Schweckendiek, 2-flap, 3-flap (V-to-Y), and double reverse z-plasty (Furlow) palatoplasties. Although most of the repairs do not involve repairing the muscular sling, doing so allows better palatal and eustachian tube functions. Descriptions of the major techniques used for palatoplasty are outlined below.

von Langenbeck technique

First described in 1861, the von Langenbeck technique underscores the importance of separating the oral and nasal cavities. Virtually every repair performed today incorporates principles initially included in this technique. Bipedicle mucoperiosteal flaps of both the hard palate and the soft palate are used to repair the defect. After their elevation, the flaps are advanced medially to close the palatal cleft. Advantages of this technique include less dissection and its simplicity. A disadvantage of the von Langenbeck repair is that it does not increase the length of the palate, which results in an inability to close primary and secondary clefts. Other criticisms of this technique include the occurrence of anterior fistulas and the resultant inferior speech due to the short soft palate. Airway obstruction during sleep seems to be an insignificant problem with this repair. Because of the physical limitations in lengthening the palate with this technique, many modifications have been made over the

years.

Schweckendiek technique

In the 1950s, Schweckendiek began to repair clefts in a staged fashion. In this technique, the soft palate is first repaired when the patient is young (typically 3-4 mo), and this is followed with hard palate closure when they are nearly 18 months. In the interim, an obturator is used to allow swallowing and speech. This technique has the advantages of achieving closure when the patients are young and causing minimal disturbance of facial growth. However, the disadvantages include the need for additional operations; the resultant speech

disorders that cannot be easily managed; and the need for frequent changing of the dental prosthesis, which can be expensive.

As noted previously, the initial repair is usually performed in a patient aged 3-12 months. The second stage is usually performed when the patient is 18 months, but it may be delayed until the patient is 4-5 years. Longer delays (ie, until primary dentition is established) were believed to be advantageous in that they prevented lateral contraction of the palatal arch. Currently, speech and feeding difficulties with delayed closure are thought to outweigh the dental alignment problems, and the current trend is to use earlier closure. Collapse of the maxillary arch is now dealt with by means of palatal expansion when the patient is young.

The initial repair is accomplished by making incisions in the soft palate along the margins of the cleft. The levator muscle, which is abnormally attached to the posterior free edge of the bony palate, is dissected free and reoriented. A 3-layer closure of the nasal mucosa, the levator muscles, and the oral mucosa is then performed. The resultant hard palate fistula is closed at a later date.

Although many methods to close the hard palate exist, one technique is the use of the vomer flap. The mucoperiosteum of the vomer bone is elevated in an inferior-to-superior direction. This flap is then rotated laterally for attachment to a small palatal mucoperiosteal flap. This procedure can provide a watertight closure with minimal elevation of the palatal mucoperiosteum. The preferred method involves raising the mucoperiosteal flaps on the oral and nasal surfaces of the hard palate and closing them in 2 layers across the defect. The vomer flap is primarily useful with wide or bilateral clefts. Vomer flaps have the disadvantage of requiring closure of 2 suture lines on the nasal surface. When used with oral mucoperiosteal flaps, the vomer flaps are attached to the flaps raised from the nasal surface of the cleft.

Two-flap technique

The 2-flap technique involves 2 posteriorly based flaps that extend the length of the defect. The flaps are rotated medially to close the defect. This method is the most common technique used for closing complete clefts. No additional length is available for closure of any alveolar defect with this type of repair. An advantage of this method is that the incidence of posterior fistula is low.

After incisions along the cleft margins have been made, the levator veli palatini muscles are dissected away from the hard palate. Modifications that include infracture of the hamulus or stripping the levator veli palatini muscle from the hamulus can be made; these changes greatly improve medial rotation of the mucoperiosteal flaps. This maneuver also reduces closure tension at the junction of the hard and soft palates, helping to prevent fistula formation. Once the nasal mucosa is freed from the nasal surface of the hard palate, the palate can be

closed in layers: the nasal and oral layers anteriorly and the nasal, muscular, and oral layers posteriorly.

Three-flap/V-Y (Wardill-Kilner-Veau) technique

In 1937, Kilner and Wardill independently described the V-Y repositioning technique. This technique is primarily used for repair of incomplete clefts or clefts of the secondary palate. The incisive foramen is the anterior border of the repair, and the uvula is completely divided posteriorly. The theoretical advantage of this technique is that pushing back the flaps adds length to the palate. This length is difficult to achieve without incising the nasal layer of the repair.

Incisions are made along the free margins of the cleft and extended anteriorly from the apex of the cleft to where the canine teeth erupt. Dissection is then continued posteriorly along the oral side of the alveolar ridge to the retromolar trigone. Mucoperiosteal flaps are elevated from the nasal and oral surfaces of the bony palate. Dissection of the greater palatine vessels from the foramen lengthens the pedicle. In the event that the vessel is avulsed and injured, the collateral flow from the lesser palatine and posterior nasal septal arteries is usually sufficient. The bony foramen surrounding the vessel can be opened posteriorly to gain more length. The tensor veli palatini muscle is elevated off the hamulus to aid in relaxing the midline closure. Billroth had advocated infracture of the hamulus, but further study revealed that this structure returned to the prefracture position within 6 months.

As in other repairs, the nasal mucosa is freed from the bony palate and closed to either side or, if necessary, closed by using vomer flaps. The muscle and oral mucosa are closed in a second single layer, usually in a horizontal fashion. Anteriorly, the oral mucoperiosteal flaps are attached to the third flap, which is the mucosa overlying the primary palate. Posteriorly, the palate is closed in 3 layers: nasal mucosa, levator muscle (which was previously freed from the bony palate), and oral mucosa.

Double reverse z-plasty

In 1986, Furlow described a technique to lengthen the velum and to create a functioning levator muscle sling. This method is difficult to perform in wide clefts. However, it is considered a good method when the cleft is narrow or if a submucous cleft exists. The technique involves opposing z-plasties of the mucosa and the musculature of the soft palate. The goal is to separate the nonfunctioning attachments to the posterior border of the hard palate and to displace the mucosa and the musculature posteriorly.

The first z-plasty is created on the oral mucosa side, while the second z-plasty is inverted on the nasal mucosa side. The incisions are made, and the oral mucosa

is dissected free from the underlying muscle. On the left side of the patient, the oral mucosa flap also contains the muscle. On the patient's right side, the muscle is kept with the underlying nasal mucosa. The 2 muscle-bearing flaps transpose posteriorly, while the thin nonmuscular flaps are placed anteriorly. This technique has the effect of rotating the muscular sling posteriorly and lengthening the soft palate. One potential problem with this technique is the formation of a fistula at the junction of the hard and soft palates.

Submucous clefts

In 1825, Roux described the most common posterior cleft: the submucous cleft. He stated that 3 factors were involved in this deformity: (1) the membranous portion of the soft palate is absent, (2) the palate is short, and (3) the nasopharynx is abnormally expansive. In 1930, Dorrance found that the anatomical defect occurred with the anatomical position of the levator veli palatini. In 1956, Calnan described the classic triad of the submucous cleft palate, defined by a bifid uvula, a palatal muscle diastasis, and a notch in the posterior surface of the hard palate.

Controversy exists regarding whether the incidence of otitis media with effusion is increased in children with submucous clefts. Studies have revealed an improvement in effusions following repair of a submucous cleft. However, more recent studies have revealed no improved resolution of the effusion after surgery.

The degree of velopharyngeal insufficiency that can exist is based on the anterior displacement of the muscles. Surgery is indicated for patients with 2 categories of conditions: (1) an overt cleft of the soft palate with velopharyngeal insufficiency or (2) an overt defect, usually undetected at birth, with a presentation of hypernasal speech. Occasionally, a submucous cleft palate defect is discovered at the time of adenotonsillectomy, either intraoperatively or postoperatively, as a complication with hypernasal speech.

Techniques for closure of submucous clefts are the same as those described above. Alternatively, the surgeon can use a pharyngeal flap technique or a pharyngoplasty. Pharyngeal flaps are usually superiorly based pedicle flaps of mucosa and underlying constrictor muscle. The overall goal is to create lateral ports that can easily close. Use of a pharyngeal flap is best when a sagittal closure pattern exists (ie, when the greatest contribution to velar closure is lateral wall movement). A sagittal closure pattern most commonly occurs with a cleft palate.

Pharyngoplasty involves 2 flaps that are positioned on either side of the pharynx and rotated superiorly to create a smaller velar opening, thereby aiding in closure of the soft palate. This method is preferred when a circular or coronal closure pattern exists because it does not interfere with the posterior motion of

the palate. The choice of technique depends on the preoperative velar closure pattern.

Alveolar bone grafting

Alveolar bone grafting is an integral part of repairing clefts that involve the anterior maxilla. Establishing a bony union can help to prevent maxillary segmental collapse, to close oronasal fistulas, and to encourage eruption of teeth. Regardless of whether the repair is early or late, the neonate should be fitted with an obturator within the first month after birth. Bone grafting in patients younger than 2 years is considered primary, and secondary grafting occurs afterward. Graft material can be obtained from the hip, the ribs, the extremities, or the outer table of the skull. Although morbidity can exist at the various donor sites, the benefit of closing the maxillary gap outweighs the potential risk.

The surgical procedure involves raising mucosal pedicles on either side of the maxillary defect. With the use of any of the described donor sites, the graft of cancellous bone is placed into the pocket. The mucosal flaps are closed in a simple fashion. Many times, the depression of the alar base is immediately corrected on completion of the procedure.

Postoperative Details

Immediate postoperative concerns in cleft palate repair include airway management and analgesia. Repairing the palate changes the nasal/oral airway dynamics and may present problems in the immediate postoperative period, especially in children with a Pierre Robin sequence. The lasting effect of narcotics used for anesthesia may also alter upper airway dynamics. Since placement of an oral airway may disrupt the palate repair, a ligature of 2-0 chromic (or silk) suture is placed through the anterior tongue to allow forward traction on the tongue while the patient is in the postanesthesia area. This suture is removed once the child is fully alert and able to maintain the upper airway.

Adequate analgesia is important in the postoperative period to allow patients to return to their activities as quickly as possible. However, the use of analgesics must be balanced with the risks of oversedation and subsequent airway compromise. Generally, acetaminophen with codeine is sufficient for this purpose. Analgesics may be continued as needed for as long as 7-10 days postoperatively with few problems; the most common adverse effect is constipation.

In infants and younger children, arm restraints or "no-no's" are used when the child is unattended to prevent the placement of fingers in the mouth because this may disrupt the repair.

Diet in the postoperative period is generally limited to liquids and soft foods that do not require chewing. The use of bottles is avoided because the nipples may

interfere with the repair. The use of spoons is also avoided for similar reasons. Feeding is accomplished by using either a cup (not a sipping cup) or a Breck feeder (a red rubber catheter attached to a syringe). Normal diet and feeding may be resumed after 10-14 days, depending on the type of repair. At 3 weeks, all dietary and feeding restrictions are removed.

Oral hygiene is best performed by rinsing with clean water, with the patient taking care to remove all collected food particles. The use of hydrogen peroxide should be avoided because it may inhibit healing. After 5-7 days, careful toothbrushing may be resumed.

Follow-up

Once discharged from the hospital, the patient should have follow-up visits at 7-10 days and at 3 weeks. If a small fistula or a wound breakdown is noted in this period, waiting at least 6 months prior to attempting closure is advised. This delay allows for maximal wound contracture and for reestablishment of the blood supply to the tissues.

Complications

Airway obstruction

As mentioned previously, postoperative airway obstruction is the most important complication in the immediate postoperative period. This situation commonly results from prolapse of the tongue into the oropharynx while the patient remains sedated from anesthetics. Intraoperative placement of a tongue traction suture helps in the management of this situation. Airway obstruction can also be a protracted problem because of changes in airway dynamics, especially those in children with a small mandible. In some instances, placement and maintenance of a tracheotomy is necessary until palate repair is complete.

Bleeding

Intraoperative hemorrhage is a potential complication. Because of the rich blood supply to the palate, significant bleeding requiring transfusion can occur. This can be dangerous in infants, in whom total blood volume is low. Preoperative assessment of the hemoglobin level and the platelet count is important. Injection of epinephrine prior to palate incision and intraoperative use of oxymetazoline hydrochloride–soaked packing material can reduce blood loss. To prevent postoperative blood loss, demucosalized areas of the palate should be packed with Avitene or a similar hemostatic agent.

Palatal fistula

Wound dehiscence (palatal fistula) can occur as a complication in the immediate postoperative period, or it can be a delayed problem. A palatal fistula can occur anywhere along the original cleft site. The incidence has been reported to be as

high as 34%, and the severity of the original cleft has been shown to correlate with the risk of fistula occurrence. Complete dehiscence is uncommon, but immediate reclosure should be attempted if it does occur. Small fistulas that occur at areas of maximal wound tension are more common. These typically occur at the junction of the primary and secondary palates anteriorly or at the junction of the hard and soft palates posteriorly.

Postoperative cleft palate fistulas can be managed in 2 ways. In a patient without any symptoms, a dental prosthesis can be used to close the defect with good results. A patient with symptoms may require surgery. Poor blood supply, especially the anterior supply, is the major reason for failure of fistula closure. Therefore, closure of persistent anterior or posterior fistulas should be attempted no sooner than 6-12 months after surgery, when the blood supply has had an opportunity to reestablish itself. Currently, many centers wait until the patient is older (at least 10 y) before attempting fistula repair. If simple closure methods fail, vascularized tissue flaps, such as an anterior tongue flap, may be required for closure.

Midface abnormalities

Cleft palate treatments at some institutions have focused on early surgical intervention. One of the negative effects can be maxillary growth restriction in a certain percentage of patients. Palates that are repaired at an early age may have a decreased anterior or posterior dimension, a narrower dental arch, or an abnormal height. Great controversy exists on this topic because the cause of the hypoplasia, whether it is the repair or the effect of the cleft itself on the primary and secondary growth centers in the mid face, is unclear. As many as 25% of patients with a repaired unilateral cleft palate may need orthognathic surgery. LeFort I osteotomies can be used to correct the midface hypoplasia, which results in malocclusion and jaw deformity.

Congenital Syndromes

Crouzon, Apert, Pfeiffer, Saethre-Chotzen, and Carpenter Syndromes. This collection of syndromes has proved to be an exciting area of investigation for plastic surgeons and other researchers. Surgeons currently have better tools to diagnose and investigate these syndromes, and they are now better understood. Most exciting of all, more sophisticated treatment methods have evolved over the past 20 years.

Crouzon Syndrome. Crouzon syndrome was first described in 1912.

Inheritance. Inheritance is autosomal dominant with virtually complete penetrance. It is caused by multiple mutations of the fibroblast growth factor receptor 2 gene, *FGFR2* (Gorry, 1995; Steinberger, 1995).

Features. Features of the skull are variable. The skull may have associated brachycephaly, trigonocephaly, or oxycephaly. These occur with premature fusion of sagittal, metopic, or coronal sutures, with the coronal sutures being the most common. In addition, combinations of these deformities may be seen (Kreiborg, 1982). The orbits are shallow with resulting exorbitism, which is due to anterior positioning of the greater wing of the sphenoid. The middle cranial fossa is displaced anteriorly and inferiorly, which further shortens the orbit anteroposteriorly. The maxilla is foreshortened, causing reduction of the orbit anteroposteriorly. All these changes result in considerable reduction of orbital volume and resultant significant exorbitism. In severe cases, the lids may not close completely. The maxilla is hypoplastic in all dimensions and is retruded. This decreases the anteroposterior length of the orbital floor. The upper dental arch is narrowed and retruded, which yields a class III malocclusion. Premature contact of the molars also may be present, resulting in an anterior open bite. This may cause the mandible to be rotated downward and backward. The chin and malars are hypoplastic.

Investigations. Examination of the eyes by an ophthalmologist is essential to assess for papilledema, which indicates elevated intracranial pressure. Another finding may be optic atrophy; fortunately, this is rare.

Radiological examination. This consists of standard radiology to produce anteroposterior, lateral, and cephalometric views. The information gained is the position of the maxilla relative to the mandible. This is class III, with the upper teeth lying behind the lower teeth when they are in occlusion. Patients show evidence of elevated intracranial pressure and have "paw marking" of the skull due to the gyri of the brain indenting and thinning the calvaria, with, in severe cases, erosion. The fusion of the involved sutures can be seen.

A CT scan helps confirm the findings of standard radiographs and provides information on ventricular size. Three-dimensional CT scans can be produced but yield no more information than standard scans, although suture fusion can be graphically displayed.

General assessment. Other abnormalities are sought, and the child's mental development is carefully assessed. An orthodontist should see the child and initiate treatment when indicated.

***Apert Syndrome*.** In nearly all patients with Apert syndrome, the cause is 1 of 2 *FGFR2* mutations involving amino acids (Ser252Trp, Pro253Arg). The condition is inherited in an autosomal dominant mode.

Craniosynostosis is present, characterized by brachycephaly and, frequently, turricephaly; the anterior fontanelle is enlarged (Kreiborg, 1991). The maxilla is hypoplastic with a high-arched palate, class III malocclusion with an anterior open bite, and, frequently, a cleft of the soft palate. The mid face is hypoplastic. Together with the retrusion, this causes exorbitism. Complex syndactyly of the hands and feet is present. It is symmetric, and other limb anomalies (eg, shortening) may be observed. The syndactyly may show fusion of the second and forth fingernails, which also may be seen in the toes (Green, 1982). Upper eyelid ptosis with an antimongoloid slant may be seen. Blindness may be present. Overall, the deformity is worse than that of Crouzon syndrome.

Pfeiffer Syndrome. This is an autosomal dominant condition caused by a single recurring mutation (Pro252Arg) of the *FGFR1* gene and several mutations involving *FGFR2*. Patients have craniosynostosis, enlarged thumbs and great toes, and a hypoplastic mid face. The hypoplastic mid face gives the forehead an enlarged appearance. The nose is small. Exorbitism may be present, but it is never as prominent as in persons with Crouzon or Apert syndrome. The condition has been classified into 3 types. Patients with type I have the best long-term prognosis, whereas those with types II and III have neurologic compromise and die young (Cohen, 1993).

Saethre-Chotzen Syndrome. This is an autosomal dominant condition with full penetrance. It is caused by multiple mutations of *FGFR2*. Craniosynostosis is present, and the hairline is low. Ptosis and brachydactyly are characteristic. The forehead is retruded, giving the appearance of slight exorbitism. The maxilla may or may not be retruded.

Carpenter Syndrome. Patients with this autosomal recessive condition have craniosynostosis, syndactyly of the feet, and short hands and fingers with syndactyly of varying degrees.

Treatment of Craniosynostosis Syndromes. The age at presentation determines the treatment. If possible, provide treatment early and direct it at the cranial vault. The aim is to reduce intracranial pressure, if present, and to prevent visual problems. In addition, the patient's appearance may be improved. The anterior cranial fossa enlargement is a result of frontal lobe growth; at 11 months, the frontal lobes are almost 50% of adult size. The anterior cranial base is at 56% of its total growth at birth and, at 2 years, has achieved 70% of its total growth, probably soon after this full growth is attained. The size and position of the anterior and middle cranial fossa floors are determined by the frontal and temporal lobes. If the anteroposterior growth of the skull base is diminished, this has no affect on mandibular growth.

Early skull base suture fusion results in interference with forward facial growth; thus, release of the affected sutures should provide normal growth. This

certainly is the case for the skull, but the mid face remains intruded in patients with Apert or Crouzon syndrome.

Undoubtedly, the best results are achieved in patients with isolated sagittal craniosynostosis. In the past, this was managed with a sagittal strip craniotomy. Currently, making coronal and lambdoid cuts to achieve immediate lateral expansion by hinging the cranial segments outwards is more common. Do not interfere with the lambdoid and coronal sutures.

At 9-11 months in persons with bilateral coronal craniosynostosis, as is seen in Crouzon or Apert syndrome, the frontal area and supraorbital rims are osteotomized separately and advanced to produce a degree of overcorrection with improved frontal contour (Posnick, 1992). This also requires a cut through the orbital roofs and in front of the cribriform plate anteriorly. Laterally in the temporal fossa, the supraorbital rim is advanced using tongue-and-groove techniques for stabilization, although this is now not absolutely necessary because plates and screws (metal or absorbable) are available. The frontal bone is now advanced and plated onto the newly positioned supraorbital rim.

Even with large advancements, scalp closure is always possible. Occasionally, this is aided by galeal scoring. Although this is a standard technique, it is modified as required by the basic anatomy. The patient is monitored using postoperative CT scans to follow the resolution of the dead space.

Distraction. Currently, the trend is to advance and fix the frontosupraorbital region and to advance the maxilla subsequently or at the same time, even at an early age, with (Chin, 1997) or without (David, 1990) a distraction apparatus. Although internal distraction devices have been used to push the maxilla forward, with fixation on the temporal area and the use of a rod to apply an anterior pushing on the lateral orbital walls, these have not been uniformly successful. This is mainly because of design faults.

The rigid external distraction device, which is secured by skull fixation, with an anterior pulling force being applied to the maxilla, is more mechanically sound and works well (Figueroa, 1999). It is expensive, the fixation can penetrate the cranium (should the patient fall, striking the side of the head), and the device can become displaced (Rieger, 2001).

Despite this, good forward maxillary movement is obtained in a relatively controlled fashion. The children do not complain about wearing this device. It can be used for persons with Crouzon or Apert syndrome or other conditions that require an osteotomy and forward pull. Whether this is an advantage over the older, well-established, and safe method of advancement osteotomy, stabilization, and bone grafting in one stage remains unproved. In older patients, the latter method probably remains the technique of choice.

Pierre Robin Syndrome. This condition consists of micrognathia, glossoptosis, and, frequently, a cleft of the palate. Airway obstruction occurs in the more severe cases. The mandible is retrognathic, and the genioglossus muscles are shortened; thus, the tongue falls back and occludes the airway. The degree of this latter anatomic anomaly varies, but it is always present and can be lethal.

The deformity may be associated with the position of the baby in the uterus, with its head flexed for a longer period than normal. In some patients, mandibular growth after birth is good, in contrast to those who remain significantly micrognathic. The child has a birdlike face with varying retrognathia and respiratory problems. He or she also may have feeding difficulties. Respiratory obstruction is present because the tongue falls back into the pharynx. The intercostal spaces retract, and the accessory muscles of respiration are overactive. The baby tends to choke, and cyanosis can be ever-present or intermittent. This occurs especially during sleep.

Feeding difficulties. These are caused by a combination of the small mandible, the position of the tongue, respiratory distress, and cleft palate. The child only takes small amounts, coughs frequently because food is inhaled, and brings much of the food back up; tracheostomy and gauge feeding may be necessary. Associated with this are malnutrition, failure to gain weight, frequent chest infections, and exhaustion. In the presence of a complete cleft, the tongue may cause respiratory problems by going through the cleft and into the nose. The large appearance of the tongue is probably relative; the mandible is small. Other airway problems should be sought, including tracheoesophageal fistula and choanal atresia. The child should be checked for other conditions that can be associated, such as cardiac, limb, and eye problems.

Treatment. The condition can range from minor to severe; therefore, treatment varies (Douglas, 1956; Moyson, 1961; Freed, 1988; Argamaso, 1992; LeBlanc, 1992; Caouette-Laberge, 1994). Mild cases are treated by placing the baby on its side or face down with the foot of the cot raised. This allows the tongue and jaw to fall into a better position and may be totally adequate. This placement allows the child to sleep, which is vital. The condition improves as the child becomes bigger and stronger.

If this maneuver is unsuccessful, the next step is intubation. This can be difficult and should be performed with the child awake. The tongue can also be advanced using a suture passed through it. It can be sutured to the lip mucosa or a Kirschner wire can be passed through its base. In this situation, the child must be watched carefully by a trained staff member. An oxygen monitor should be placed in addition to a cardiac monitor.

If the child continues to need an endotracheal tube after 2-3 days or cannot control its airway after a week of conservative treatment, consider surgery. A

tracheostomy can be difficult in these small children; if possible, a Björk flap should be used. This allows for greater ease of changing endotracheal tubes and maintains patency of the tracheostome.

Suturing the tongue to the lower lip to open the airway has become more popular. This can deform the lip and tongue, but it may be effective for opening the airway. Other procedures involve fixation of the tongue in a forward position using a Kirschner wire and many other variations of the tongue-to-lip adhesion.

The introduction of mandibular distraction has yielded the possibility of immediate correction of the deformity by lengthening the mandibular body bilaterally (Molina, 1995). The distractor is applied to the body of the mandible, and a partial body osteotomy with preservation of the mandibular nerve is performed. The distractor may be external or internal; the latter is almost becoming standard in the author's practice. It is easy to perform and is simple for the parents to manage. The procedure is performed bilaterally, and the mandibular body is lengthened; thus, the tongue is brought forward. A period of consolidation occurs once a slight overcorrection has been achieved. The external method is used in many centers and is equally effective. This is a logical method that corrects the primary deformity. The other methods have been less effective and have resulted in deformity.

Predicting how these children will fare in the long term is always difficult. Some develop normally, and others may require further mandibular surgery to achieve an acceptable facial profile. Proper care by experienced surgeons in good centers has greatly improved the outlook for these children. Most should survive if the proper decisions are made at the correct times. Management by experienced surgeons, anesthesiologists, and nurses is essential

Hemifacial Deformity. *Hemicraniofacial Microsomia.* Poswillo has suggested that this condition results from bleeding in the temporal area in the embryo. It has also been associated with thalidomide intake during pregnancy. If epibulbar dermoids and vertebral anomalies are present, it is termed Goldenhar syndrome. This condition can be bilateral. The components of the syndrome are low-set ears, short mandibular ramus, micrognathia, chin deviation, occlusal cant, and anterior open bite.

The zygomatic arch may be present, absent, hypoplastic, or partially absent. Severe deficiency of the subcutaneous soft tissue and absence of temporal hair may be observed. The orbit may be displaced, deformed, and small. The eyes may be rudimentary or absent, and the lateral canthus can be displaced. The side of the face may be hypoplastic or vertically short. All degrees and variations of this deformity may occur. Many classifications schemes have been developed, but most are unhelpful.

A careful assessment of both the hard and soft tissue deficiencies is advised, while keeping treatment considerations foremost in mind. The major features can be considered as auricular, maxillary, and mandibular underdevelopment. In severe cases, the skull is also involved, especially the sphenoid and temporal bones.

The mandibular deformity can range from (1) minimal hypoplasia to (2) a small ascending ramus and condyle with an absent glenoid fossa and, possibly an absent coronoid to (3) a minimal or absent ramus with severe vertical facial shortening.

These deformities become worse with time. Flattening and hypoplasia of the zygomas and its arch (the latter may be absent) also may occur. The muscles of mastication (ie, masseter, medial and lateral pterygoids, temporalis) may be hypoplastic. This upsets the functional matrix, which further affects related bony development.

Ear deformities vary in severity. Grade 1 is small, malformed ears. Grade 2 is a vertical remnant of skin and cartilage with no ear canal. Grade 3 is a small remnant and lobule. Patients also have variable degrees of deafness. Audiometry and CT scanning are imperative. Look for possible cerebral anomalies.

Cranial nerves. Abnormalities are common in patients with craniofacial microsomia. The facial nerve is most commonly affected to a greater or lesser degree. Optic, trigeminal, cochlear, and cranial nerves are involved.

Facial clefts The distance between the eye and the angle of the mouth is shortened. Transverse facial clefts are seen and vary in severity. They span the angle of the mouth to the ear.

Renal. The patient may have a hypoplastic or missing kidney.

Investigations. Panorex, cephalograms, CT scans, 3-dimensional CT scans, and MRI are all helpful in the assessment of the deformity. Dental impressions are taken.

Treatment. Treatment is long-term. Unilateral or bilateral mandibular distraction can be performed early, especially in patients with bilateral deformity and airway problems. The skull deformity and orbital deformity can be corrected before the child starts school. Skull grafts can be used to reconstruct the orbit, zygomatic arch, and malar area when the ascending ramus of the mandible requires reconstruction. A costochondral graft can be effective. Later, a Le Fort I and sagittal split mandibular osteotomy is performed with chin advancement as necessary. When an ascending ramus is required, a rib graft can be effective. Soft tissue is supplied by free tissue transfer.

The facial cleft is repaired in layers. As in the cleft lip, the muscle layer, frequently disregarded in the past, is most important.

Orthodontic supervision is essential, and further maxillary and mandibular osteotomies are frequently necessary when the face is mature (age >16 y).

Even with all these well-planned procedures performed as indicated over the years, the end result may leave much to be desired.

Romberg Disease (Hemifacial Atrophy). This is a sporadic condition in which progressive atrophy of skin, subcutaneous fat, muscle, and bone occurs. The contralateral face may be involved, but this is uncommon. The process can go on for a variable number of years. The skin and adnexa atrophy, fat becomes inflamed, and muscle atrophies. Nerves are unaffected. An association with sympathetic stimulation has been postulated, but the cause remains unknown.

Differential diagnosis. Lipodystrophy produces a similar appearance, but only in fat and is often bilateral. Scleroderma is similar to hemifacial atrophy.

Treatment. Treatment should be delayed until the process has ended.

Bone. This can be augmented by iliac crest or split skull, the latter being taken from the contralateral side. Fixation should be rigid. The skull can be also augmented with methyl methacrylate or hydroxyapatite cement (Bone Source, Stryker Leibinger, Freiburg, Germany; Mimix, Biomet, Warsaw, Ind; or Norian CRS, Synthes-Stratec, Oberdorf, Switzerland).

Soft tissue. This should be replaced via transfer of vascularized free tissue, deepithelialized skin, subcutaneous tissue, and fat. Muscle can be incorporated. The use of muscle alone is not recommended because it tends to atrophy over time. Occasionally, a skin island is be used to release and augment the skin area. The free tissue transfer should be securely fixed, preferably to bone with nonabsorbable sutures to avoid slippage. Omentum can be used, but it tends to sag even with secure and plentiful fixation. Pedicled galea is available but is limited to cheek defects of small volume and is not always reliable. Tongue flaps can provide filling material for atrophic lips. Dermis can also be used, but this frequently loses considerable volume. Rarely is total symmetry achieved.

Klippel-Feil Syndrome. This condition has 3 radiologic types. The first is characterized by continuity of cervical and upper thoracic vertebrae. The second is characterized by involvement of only a few interspaces but with hemivertebrae with scoliosis and fusion of the atlas to the skull base. The third is characterized by fusion of the cervical, lower thoracic, and lumbar vertebrae (Erskine, 1946; Guille, 1995).

Clinical features. Patients have a short neck, restriction of neck movement (especially rotation), and a low occipital hairline. Flexion and extension is better

than rotation. Associated features are webbing of the neck, scoliosis, torticollis, and, occasionally, Sprengel deformity, which is displacement of one or both scapulae. In some cases, patients have mental retardation. The patient also may have problems with eye movement, hearing loss, and urinary anomalies. One third of patients have unilateral renal agenesis, and vaginal agenesis has also been noted in a few children.

The syndrome occurs in 1 in 42,000 births and is more frequent in females than males. Type I is most common clinically, and type II causes fewer clinical problems and is often found only at autopsy. Palatal clefts alone tend to occur 3 times more frequently than combined lip and palate clefts. Cardiac anomalies, especially ventricular septal defects, are more common in affected children than in healthy children.

Other syndromes with similar cervical findings are Noonan and Morquio syndromes; spinal tuberculosis can have a similar involvement. The cause is unknown.

Cause. The process is failure of segmentation of the cervical spine. The cause of this has been postulated to be a lack of midline fusion with disruption of the notochord. A second theory is that the subclavian artery has been disrupted.

Treatment. This depends on the extent of the problem, which may consist of cardiac, palatal, and skeletal involvement. If cervical stability is in question, anesthesia may be a problem. It must be performed with care to prevent any neurologic problems. Spinal fusion should be performed, and cord pressure should be decompressed. Correction of neck webbing is difficult and consists of resection of excess skin and underlying soft tissue with Z-plasty skin closure. The platysma rather than the deep muscles are involved. Other types of skin plasties have been described, but all produce unsatisfactory scars. The posterior hairline is corrected by expansion of the neck skin with subsequent excision of the hair-bearing area with expanded skin advancement.

Turner Syndrome. This occurs in females. A webbed neck, infantilism, and cubitus valgus are observed. Another term for this condition is pterygium colli. Patients have ovarian agenesis and dwarfism. These girls have only 44 autosomes and one sex chromosome, the X-O.

The findings are dwarfism, webbed neck, low hairline, epicanthal folds, mandibular deformity, webbed elbows and knees, coarctation of the aorta, hypertension, lymphedema of the hands and feet, and mental retardation. A buccal smear is taken, and the absence of a peripheral chromatin mass confirms the diagnosis.

Treatment. A gynecologist should treat these patients in conjunction with an endocrinologist. Webbing of the neck can be corrected with an unequal Z-plasty.

Hair is excised from the lateral hair-bearing area, and a small hair-bearing flap is transposed into a lateral defect resulting from a large flap being taken and placed posteriorly to raise the posterior hairline. Another method, as mentioned earlier, is to expand non–hair-bearing skin and advance this after excision of the posterior hairline. If only the lateral web needs to be addressed, multiple Z-plasties may be used; however, the scars can be obvious and may be of poor quality.

Various midline resections can be made on the back of the neck, with advancement of the skin edges in different designs, such as advancement lateral to medial with X or Y scars. Although improvement of the webbing is achieved, it is never complete and the scars (eg, keloid, hypertrophic, stretched) can be most unsatisfactory.

Treacher Collins Syndrome (Mandibulofacial Craniosynostosis). This condition is passed on as an autosomal dominant gene with variable penetrance and phenotypic expression. It is a bilateral condition that affects first and second arch structures. As with all inherited conditions, it varies from mild to severe. Patients have hypoplasia of the zygomas and mandible with ear defects or an absence and displacement of the sideburns. The eye area can be significantly involved, with an antimongoloid slant of the fissures, colobomas of the lower lids, and an absence of eyelashes medially.

The nose is beaked downwards at its tip. In severe cases, patients have thinning of the subcutaneous tissue from the angles of the mouth up to the lateral canthus. This is associated with macrostomia and is similar to a subcutaneous cleft. The texture of the skin in this area is different because of its thinness. It is also known as Franceschetti-Klein syndrome. The face is fishlike, and patients may have deafness and mental retardation.

Other associated problems include vertebral anomalies, clubfoot, lung agenesis, and frontalis agenesis. The hypoplastic mandible, glossoptosis, small size of the pharynx and nasopharynx, and occasionally choanal atresia can cause severe breathing problems and even death. A more minor situation causes sleep apnea, with all of its attendant problems.

Patients have a family history positive for the syndrome. The condition is transmitted by autosomal dominance, and the penetrance and expressivity are variable. The mother transmits the gene, and it may become more lethal from generation to generation. The syndrome seems to be more prevalent in children born to parents of older age. An increased incidence of spontaneous abortions is observed.

Differential diagnoses include Nager syndrome and ablepharon macrostomia syndrome. The syndrome may be due to problems of differentiation of the branchial arch mesoderm interfering with facial bone development. Stapedial

artery hypoplasia may cause ischemia in the facial region. A significant portion of the craniofacial area, soft tissue and cartilage, bone, and teeth are derived from the neural crest ectoderm. The gene for Treacher Collins syndrome has been mapped to band 5q31.3-q33.3 (Jabs, 1991). This has been reproduced in an animal model by damaging neural crest cells using vitamin A.

Overall management. These patients require treatment by a multidisciplinary team, including a plastic surgeon, oral surgeon, orthodontist, oculoplastic surgeon, and, occasionally, a neurosurgeon. The social worker plays an important part in the social aspect of the family.

Shortly after birth, the patient may develop airway problems; the help of an otolaryngologist may be necessary. In some cases, simple head-down positioning with elevation of the foot of the crib may be sufficient; in others, a tracheostomy is considered. More recently, as described for Pierre Robin syndrome, early bilateral mandibular distraction can be performed to correct the airway problem.

Others involved with the treatment of these children are audiologists and speech pathologists. Later, they may require a psychiatrist. Feeding difficulties may require the help of a nutritionist.

Skeletal assessment. This is essential prior to contemplating treatment. Use standard facial bone radiographs, CT scans, and 3-dimensional CT scans.

The orbital shape shows an inferolateral rotation with an absence of the floor. The malar area and zygomatic arch are missing to a variable degree. The central portion of the maxilla seems somewhat protruded and vertically shortened; it is also narrower than normal. The mandible shows an increasing deformity from hypoplasia of the glenoid fossa and ascending ramus to complete absence of these areas with a free-floating mandible. The classifications are type I, type II subtypes A and B, and type III. The facial soft tissue is also disrupted.

Treatment. The mandible can be treated early using bilateral osteotomies in the body with distraction, either intraorally or extraorally. Many different methods have been described for the orbitomaxillary reconstruction. The requirements are a recontour of the orbit and a reconstruction of the lateral orbital wall and orbital floor together with the zygomatic arch and malar region.

Many types of bone have been used in the reconstruction, including rib, split skull, vascularized cranial bone on temporalis muscle, and iliac crest bone grafts. The author has tried all of these and has developed a reconstruction algorithm.

At approximately age 5-6 years or when the patient presents to the surgeon, the problem is assessed using clinical examination and 3-dimensional CT scans. Minor deformities are augmented with cranial or iliac crest bone grafts rigidly

fixed with screws and, if necessary, plates. This can be performed from an intraoral approach. With more significant bone absence, a coronal flap exposure is used to expose the orbital and malar area. The orbital contents are radically freed from the facial soft tissues and any bony attachments so that they can be reduced into the orbit. A model of the missing bone, zygomatic arch, and malar is obtained. This is then transferred to the temporal area, and the design is marked on the skull with a pencil.

A full-thickness cranial bone graft is taken by cutting along the plan using a contouring burr or an air drill. A microplate is then contoured to form the shape of the zygomatic arch and the lateral aspect of the malar area. The bone graft is osteotomized to fit onto the shaped microplate. The latter is then screwed onto the malar bone and the small nubbin of bone in the preauricular area, which represents the rudimentary lateral origin of the arch (Posnick, 1993).

Further skull bone grafts are taken as required for the lateral orbital rim and the lateral aspect of the supraorbital rim. Prior to this, the junction of the supraorbital and lateral orbital rim is contoured to a normal shape. The bone grafts are fixed with screws. In more severe cases, a significant defect remains in the floor, particularly in the lateral area. This is also reconstructed with a bone graft, which often must be wired into position. In patients with larger lateral defects, stacked grafts are used.

Attention is now turned to the skull defect. This is filled with bone shavings taken from the skull with a sharp osteotome. From the posterior scalp, a large sheet of pericranium is raised. This is used to cover the zygomatic arch reconstruction and to smooth out the whole area. Fortunately, the upper lids have an excess of skin. After closure of the scalp, attention is turned to the eyelids. The excess of skin is taken down as a flap based laterally. The skin of the tight lateral area of the lower lid is totally released, and the upper lid flap is inserted into the defect. This allows the lateral canthus to be elevated and fixed to the inner aspect of the lateral orbital rim. Overcorrection is advised.

This procedure corrects much of the deformity; however, a residual vertical shortage of the lower lid remains laterally. This occurs because of a deficiency of the internal lamella of the lid. After 3-6 months, this is corrected. Recently, a newly described method of internal lamella reconstruction has been described that has, so far, yielded the best result. The lid is injected with a vasoconstrictor and is released on its inner aspect using a horizontal incision.

When the correct height is obtained, a shaped conchal cartilage graft, with intact perichondrium on the surface facing the eye, is sutured into place with buried sutures. The perichondrium epithelializes, and a good result is usually achieved. In the past, a full-thickness upper lid island flap was used, but the present

method has been found to be more suitable. If lateral canthal adjustment is required, this can be performed concurrently.

The mandibular retrusion can now be managed using distraction osteogenesis (McCarthy, 1992). Complex cases require glenoid fossa and perhaps ascending ramus reconstruction. Costochondral grafts are occasionally required for ascending ramus reconstruction. The author has found that temporal skull augmentation is frequently required later. This is performed using alloplastic material, methyl methacrylate, or one of the hydroxyapatite pastes.

Later reconstruction. In adolescence, after orthodontic management, a bimaxillary procedure is performed. Usually, this consists of a Le Fort I and a sagittal split advancement of the mandible with an advancement genioplasty.

The author typically performs the necessary upper face augmentation using a coronal flap approach. Skull bone grafts are favored for the reconstruction. At the same time, a rhinoplasty with any necessary airway surgery is performed. In some cases, nasal augmentation is required. This is accomplished with a split- or full-thickness cranial bone graft as required. Occasionally, a repeat lateral canthopexy is required. In all of these cases, remember that the problem with the airway remains regardless of the patient's appearance; consequently, all of the precautions necessary in such a situation must be observed.

These patients have greatly improved appearance; thus, their confidence increases. The beneficial results of the surgery bring both increased job opportunities and better chances to meet others, which leads to a more normal social life. One problem is that the Treacher Collins condition may become more common as more people with the condition have children and pass the trait to their offspring.

The ear reconstruction is performed at age 6 years using costal cartilage in the manner described by Brent and others (Brent, 1992).

Hypertelorism (Greig Syndrome). In persons with this condition, the orbits and their contents are shifted laterally, either by a midline cleft or excessive ethmoid sinuses (Greig, 1924; Cohen, 1979). The anterior skull may be flat, and the frontal area may be somewhat retruded. The maxilla may be hypoplastic and/or retruded with a class III dental occlusion. Often, diastasis of the upper incisor teeth is seen. The nose may be bifid. In rare cases, the mid face and palate may be widely clefted. The condition may be asymmetric, but this is usually associated with clefts. The patient's level of intelligence may vary from normal to significantly reduced.

Treatment. Minor degrees of deformity (referred to as telecanthus) can be corrected by removing a small amount of bone in the midline (Tessier, 1967; Tessier, 1974; van der Meulen, 1979).

Osteotomies of the medial orbital walls are performed, and the walls are moved together and stabilized to the glabellar areas and to one another with wires or miniplates. The approach is by a coronal flap. Usually, one need not remove skin; however, an ellipse can be resected if indicated. In some cases, the nose is flat and may require reconstruction with a cranial bone graft. When the intercanthal distance is such that a conservative approach will not yield a good result and when a degree of downward rotation of the orbit is present laterally, a more involved operation is necessary. A coronal approach is used to explore the frontoorbital region. The temporalis muscles are dissected from the fossa.

A limited frontal craniotomy is performed in the midline. This allows a total orbital osteotomy to be performed. This is performed after the central bony block of a precalculated width is removed. The central cribriform area is removed together with the frontal sinuses. The orbits are mobilized and brought together. If necessary, the lateral orbits are rotated superiorly. This allows correction of the antimongoloid slant by superior fixation of the lateral canthal ligaments. A cranial bone graft may be necessary for correction of the flat nasal bridge line. When the hypertelorism is wide with a midface cleft or when the dental arch is distorted because of vertical maxillary shortening in the midline, a different and more satisfactory procedure termed the facial bipartition is used (Tessier, 1974; van der Meulen, 1979).

Through a coronal approach, a V-shaped osteotomy is formed from the frontal area down to the nasal bones. With a fine osteotome, an osteotomy is made between the upper incisions and the cut is continued along the hard palate. The zygomatic arch is cut, the retrotuberosity osteotomy is performed, and all walls of the orbit are osteotomized.

An osteotomy is made on the orbital roofs to meet the medial and lateral wall osteotomies. With maxillary mobilizing forceps, the 2 segments are loosened. Now they can be rotated together superiorly using the incisor osteotomy as the center of the arc of rotation. The orbits are brought together in their correct axis in 3 dimensions. This is a stable osteotomy after it is fixed. The soft tissue is reapplied, placing the lateral canthi in their correct positions. The nose may require bone grafting. If the patient has a midline facial cleft, it is repaired in layers. All bony fixation is with plates and screws.

Hypotelorism. This is a rare condition that may not be compatible with life. In the most severe cases, the infant looks like a cyclops. The anatomy is complex. In most cases, the intercanthal area is narrowed, and varying degrees of frontonasal hypoplasia are seen. The orbits are large and may be oddly shaped. Frequently, the patient has a lower-than-normal level of intelligence.

Treatment. See also Trigonocephaly - Metopic craniosynostosis. The frontonasal supraorbital area is exposed through a coronal approach. Bilateral orbital

osteotomies are performed, and the orbits are moved apart. The central defect is grafted with bone. Usually, a bone graft is used to supply a nasal bridge line. Characteristically, the nose is long and cannot be effectively shortened. The medial canthi are repositioned. When this has been achieved, a soft tissue defect is formed, which can be reconstructed using a local flap. The end result can be satisfactory, but, undoubtedly, the central portion of the face is long. Associated upper and/or lower lid colobomas may be seen. These must be repaired in layers in a standard fashion. The best plan is to correct these prior to performing the orbital repositioning.

Craniosynostosis. This condition has been known since antiquity. Hippocrates was the first to describe it in 100 CE. Others reported deformed skulls without sutures. Even in these early days, premature closure of the sutures was known to cause specific skull deformities. Under this heading, isolated craniosynostoses are compared to syndromes of craniofacial synostoses such as Apert and Crouzon syndromes, which are considered separately.

Common deformities that occur as a result of premature suture fusion are scaphocephaly, trigonocephaly, brachycephaly, plagiocephaly, oxycephaly, and turricephaly. The sutures involved in these deformities are sagittal, metopic, bicoronal, unicoronal, and multiple for the last 2 conditions. Complex premature suture fusion is associated with Apert, Crouzon, kleeblattschãdel, Pfeiffer, Saethre-Chotzen, and Carpenter syndromes. The cause of the premature fusion remains unknown.

Symptoms. In many cases of craniosynostosis, patients have no true symptoms, only deformity. Single-suture craniosynostosis rarely produces a significant rise in intracranial pressure, but 2 or more can certainly cause this (Renier, 1982; Speltz, 1997). When the increased pressure is established, the brain pushes against the skull to cause "thumb printing" or, in some cases, true erosion over the gyri. This is a definite indication for surgery. Hydrocephalus may be present and should be assessed with the placement of a ventriculoperitoneal shunt if the degree of pressure warrants such a measure (Shuster, 1995). A further complication of this situation is blindness (Tuite, 1996).

Visual deterioration is associated with elevated intracranial pressure. This is usually observed in multiple-suture stenoses (eg, oxycephaly, brachycephaly, Crouzon syndrome, Apert syndrome). It is rarely observed in plagiocephaly, scaphocephaly, and trigonocephaly. The cause of blindness is not actually known because the optic canals are of normal dimensions. It may result from increased intracranial pressure, a change in the skull base dimensions, or chronic papilledema. In patients with severe hypertelorism, their wide-set eyes may prevent them from developing binocular vision.

Craniofacial development. Knowledge of the time scale of craniofacial growth is important. The anterior cranial fossa enlarges as the frontal lobes enlarge with growth. They are 47% of adult size at 11 months and 93% of adult size at age 7 years. By age 2 years, the anterior portion of the cranial base is at 70% of its total growth. The situation with the temporal lobes is somewhat similar. The nasomaxillary complex follows the growth of the anterior skull base.

Plagiocephaly - Unilateral coronal craniosynostosis. When one coronal suture fuses prematurely, the frontal area is flat, the supraorbital rim and the lateral orbital wall are retruded, and the orbit is narrowed transversely. Coronal synostosis is due to an amino acid substitution (Pro250Arg) that results from a single-point mutation in the *FGFR3* gene on arm 4p (Cohen, 1997).

The palpebral fissure is narrowed, and the eyebrows are elevated laterally. The nose may be deviated to the affected side, and the ear may be displaced anteriorly. Radiologically, the orbit shows the harlequin deformity due to the elevation and retrodisplacement of the lesser wing of the sphenoid (Faure, 1967). Frequently, occipital bulging is observed. The condition is rare, occurring in 1 in 0,000 live births (Roddi, 1995). This situation should not be confused with deformational unilateral skull deformity.

Treatment. Correction is performed when the patient is aged 6 months, if possible (Jackson, 1981; Marchac, 1981). Several methods are available for treating this deformity. The involved portion of frontal bone is removed, and this may be rearranged by osteotomies or by barrel-staving. In the latter, a series of parallel cuts are made through the bone until it can be reshaped. The lateral orbital wall and supraorbital rim are modified by osteotomies. When the desired shape is achieved by whatever method is chosen, this complex is replaced and stabilized by resorbable plates made from polylactate combined with polyglycolate. Titanium microplates can be used to assemble bone plates, but these are then placed onto inner-aspect segments to obviate any erosion towards the brain and to eliminate any irregularities under the scalp.

In an alternative method, the supraorbital rim is expanded by inserting a small cranial bone graft into a central osteotomy. The size of the graft is determined based on measurements of the normal supraorbital rim. The frontal area is osteotomized and rearranged as necessary. Again, plates (absorbable or metallic) are used on the intracranial surface and to fix the cranium and supraorbital rim together. Absorbable plates secure the skull and supraorbital segment in place if the whole misshapen orbito-orbitofrontal segment is totally remodeled to provide symmetry with arrangement reconstruction and fixation is performed rapidly and securely.

Deformational unilateral skull deformity. The sutures are intact. However, because of the child's position in the uterus, the head is asymmetric but the orbit

is less involved, no harlequin sign develops, no nasal deviation occurs to the ipsilateral side, and no ridge forms over the coronal suture. Other causes include the practice of placing sleeping infants on their backs in order to reduce the risk of sudden infant death syndrome (Turk, 1996) and a greater number of infants being referred for early assessment (Huang, 1996).

It occurs in 5-25% of babies (Kane, 1996; Persing, 1997). Some degree of occipital flattening occurs. When the skull is viewed from above, the ear is markedly displaced backwards on the flattened side. Usually, this condition is self-correcting; however, occasionally, surgical correction is indicated (Hansen, 1994). Helmet treatment can also be used, but ideally, this should occur within the first 6 months of life (Clarren, 1981).

Differentiation. On CT scan images, especially 3-dimensional CT scan images, the lambdoid suture is open. Posterior flattening occurs along with contralateral anterior ipsilateral flattening. The ear on the affected side is displaced anteriorly when viewed from above.

Treatment. Surgical treatment is necessary for unilateral craniosynostosis or severe postural deformity. If the orbit is constricted, it is enlarged by a vertical osteotomy in the supraorbital rim and a bone graft is placed. The coronal osteotomy is rotated to place the flattened area under the hair-bearing scalp on the noninvolved side. A lateral canthopexy is required, and, rarely, a medial canthopexy must be performed (Jackson, 1981). Consider helmet treatment in less severe cases (Clarren, 1981).

Bilateral coronal craniosynostosis. Both coronal sutures are fused, resulting in a widened head with a decrease in anteroposterior dimensions. The supraorbital rim is recessed, and the eyes are prominent. The face is usually unaffected. This condition can be nonsyndromic or syndromic when a component of Crouzon, Apert, Jackson-Weiss, or Pfeiffer syndrome is present. This causes variable degrees of brachycephaly. The syndromes can be defined at a molecular level (Muenke, 1995). Pfeiffer syndrome is heterogenous with a single recurrent mutation (Pro252Arg) of the *FGFR1* gene and mutations affecting *FGFR2*. Crouzon syndrome is associated with multiple mutations of *FGFR2*. Jackson-Weiss syndrome is associated with an *FGFR2* mutation. Apert syndrome is associated with an *FGFR2* mutation.

Surgery is performed when the patient is aged approximately 6 months, but it can be performed earlier. Treatment begins with a bifrontal craniotomy. Following that, the supraorbital rims and glabellar area with the lateral orbital walls are removed en bloc. In the temporal fossa, a tongue-and-groove arrangement provides stability. Both of these segments may require some modification to make the forehead less flat. They are then fixed together as described in the previous section and are then advanced as required.

Fixation is usually performed with metal miniplates in the temporal region because the resorbable plates tend to bend when the scalp is returned. If closure becomes difficult because of the advancement, the whole scalp is mobilized posteriorly. If closure remains difficult, the galea is incised with multiple cuts from side to side. This yields satisfactory scalp expansion (Posnick, 1992). Because of their young age, these patients do not require bone grafts. Bone grafts are required in older patients, and a split-skull graft is the graft of choice.

Trigonocephaly - Metopic craniosynostosis. The midline forehead suture fusion results in a narrow anterior cranial fossa with a midline keel. Patients have hypotelorism with narrowing of the area between the medial canthi, and their orbits are rotated posterolaterally (Posnick, 1994).

Treatment. A central coronal osteotomy splits the anterior cranial fossa and the supraorbital rims. The latter are rotated anteriorly and hinged medially, as are the frontal bones. This enlarges the anterior cranial fossa. The head is widened anteriorly (Sadove, 1990).

Acrocephaly or turricephaly. This can result from multiple-suture fusion; thus, the skull grows upward. Basal suture fusion reduces the dimensions of the skull base.

Kleeblattschãdel anomaly. The characteristic of this deformity is the cloverleaf skull. The vertex bulges upward, and the bitemporal regions bulge laterally. This gives the skull the appearance of having 3 lobes. These are superior anteriorly and bilateral in the temporal area. Variable degrees of this condition are reported. In some areas, the suture is closed; in others, the sutures may be wide open. The flow of cerebrospinal fluid seems to be interrupted in the region of the third ventricle. These children have a high prevalence of mental retardation. This is also the case in children with Apert syndrome.

Treatment. This condition is treated similar to other craniosynostosis syndromes. The skull is exposed using a coronal flap, with a posterior coronal flap to widely expose the frontotemporal and occipital areas. All involved sutures are removed, and the individual areas of the skull are mobilized to adequately enlarge the skull. The deformed portions of the skull (eg, the temporal areas) are frequently recontoured by barrel-staving and molding. The bony segments are minimally stabilized to allow brain growth to proceed as normally as possible. Further releases may be required in the long term.

Sagittal craniosynostosis – Scaphocephaly. This condition produces a long, narrow skull with some rotation of the supraorbital area such that the medial area is in advance of the lateral orbital region (Hunter, 1976; Pollack, 1996).

Treatment. Two methods are available to address this situation (Kaiser, 1988). A central strip of cranium can be removed from the coronal sutures to the occipital

suture. However, if the surgeon is worried about the underlying sagittal sinus, strips of cranium can be removed from either side of the fused suture. Once this is performed, cuts are made anterior to the occipital suture and posterior to the coronal suture. The lateral skull segments are then out-fractured.

If the frontosupraorbital area is unduly deformed as described earlier, it is removed en bloc and osteotomized in the midline to establish a proper frontosupraorbital contour (Clarren, 1981). These 2 procedures combined, when indicated, achieve adequate correction of this deformity. The occiput does not require correction because it is hidden by the hair, especially in females. If significantly involved, the posterior fossa may be enlarged by central and lateral osteotomies. This should be performed with care because the lateral sinuses can be transgressed, with disastrous results.

Van der Woude Syndrome. **Mouth and facial deformities present.** Van der Woude syndrome consists of mouth and facial deformities which may vary from person to person and in degree of severity, even among members of the same family. Van der Woude syndrome affects about 1 in 100,000-200,000 persons worldwide, both males and females, and people of all ethnic backgrounds. Many people inherit the syndrome, but others have no affected relative.

Symptom sand Diagnosis The deformities present in van der Woude syndrome, which form the basis for diagnosis, typically consist of:

- A split (cleft) in the roof of the mouth (palate) and/or in the upper lip. This may occur on one or both sides of the mouth.
- Small pits in the center of the red part of the lower lip. They may appear as bumps on an infant’s lip, changing to depressions as the child grows older.

About 70% of individuals affected by van der Woude syndrome have both deformities. Some individuals have only cleft palate or only lip pits. Other less common signs of van der Woude syndrome can be:

- Missing teeth on lower jaw (incisors or premolars)
- Congenital adhesion of the jaws
- Narrow, high, arched palate
- Tongue deformity

Anomalies of other parts of the body have been reported with van der Woude syndrome, but it is not clear if these malformations are truly part of the syndrome.

Treatment. Plastic surgery can repair cleft lip and palate. Lip pits are often removed surgically. Dentists and plastic surgeons can repair other problems with the teeth or jaws.

Genetic counseling for individuals with van der Woude syndrome is important. A parent with only mild symptoms needs to know his/her child would be at risk for serious mouth and face malformations, since the syndrome's effects may vary even from parent to child. Since van der Woude syndrome is an autosomal dominant disorder, each of this parent's children would have a 50% chance of inheriting the syndrome. The gene for van der Woude syndrome has been identified on chromosome 1, but genetic testing for it is available at only a few laboratories.

Goldenhar Syndrome and Gulf War Veterans. Rare disorder may be more prevalent in their children. The data began to trickle in beginning nine months after the end of the Gulf War. Veterans who had fought in the Persian Gulf were reporting that their children were being born with birth defects, and it seemed to be at more than the rates that occur naturally by chance.

One of the rare disorders that seemed to be more common for the veterans' children was Goldenhar (pronounced golden-hair) syndrome. Medically, this is called oculo-auriculo-vertebral (OAV) spectrum. It has a wide range of symptoms and may look very different from one child to the next. However, it tends to produce:

- Face smaller on one side than the other and facial deformities
- "Oculo": Eyes abnormally small or part of upper eyelids missing
- "Auriculo": Malformation of the ears, such as being smaller than normal and with outgrowths of skin
- "Vertebral": incomplete development, fusion, or absence of certain vertebrae
- Other internal problems with heart, lung, kidneys, or intestines

Department of Defensestudy. In 1995, in response to Gulf War veterans' concerns about the unusual number of their children born with Goldenhar syndrome, the Department of Defense sponsored a study. The researchers looked at the birth records of all babies born between 1991-1993 in military hospitals after the Gulf War. The results were:

- Infants of Gulf War veterans: 5 cases of Goldenhar syndrome (rate = 14.7 per 100,000)

- Infants of veterans who did not deploy to the Persian Gulf: 2 cases (rate = 4.8 per 100,000)

The authors of the study stated, "The few affected cases . . . require these results be interpreted with caution and do not exclude chance as an explanation for these findings." Problems with the study were that it only included births in military hospitals, and did not include children who were diagnosed with Goldenhar syndrome later on.

Birth Defect Research for Children. A nonprofit group originally called Association for Birth Defect Children, now Birth Defect Research for Children (BDRC), started a National Birth Defect Registry in 1990 to collect comprehensive data on birth defects. Information is reported to the registry by parents. Using its Registry data on Gulf War veterans' children, BDRC found that:

- Between 1991-1997 there were 15 reported cases of Goldenhar syndrome among children of Gulf War veterans (includes births at both civilian and military hospitals)

One problem with the BDRC data is that the total number of births is not reported, so the actual rate can't be determined for the 6-year period involved.

Patau Syndrome (Trisomy 13). Extra copy of chromosome 13. Patau syndrome, Trisomy 13, is the least common of the autosomal trisomies, after Down syndrome (Trisomy 21) and Edwards syndrome (Trisomy 18). The extra copy of chromosome 13 in Patau syndrome causes severe neurological and heart defects which make it difficult for infants to survive. The exact incidence of Patau syndrome is not known, although it appears to affect females more than males, most likely because male fetuses do not survive until birth. Patau syndrome, like Down syndrome, is associated with increased age of the mother. It may affect individuals of all ethnic backgrounds.

Symptoms. Newborns with Patau syndrome share common physical characteristics:

- Extra fingers or toes (polydactyly)
- Deformed feet, known as rocker-bottom feet
- Neurological problems such as small head (microcephaly), failure of the brain to divide into halves during gestation (holoprosencephaly), severe mental deficiency
- Facial defects such as small eyes (microphthalmia), absent or malformed nose, cleft lip and/or cleft palate

- Heart defects (80% of individuals)
- Kidney defects

Diagnosis. The symptoms of Patau syndrome are evident at birth. Patau syndrome may be mistaken for Edwards syndrome, so genetic testing should be done to confirm the diagnosis. Imaging studies such as computed tomography (CT) or magnetic resonance imaging (MRI) should be done to look for brain, heart, and kidney defects. An ultrasound of the heart (echocardiogram) should be done given the high frequency of heart defects associated with Patau syndrome.

Treatment. Treatment of Patau syndrome focuses on the particular physical problems with which each child is born. Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help individuals with Patau syndrome reach their full developmental potential.

Genetic counseling and support. Parents of a child born with Patau syndrome will receive genetic counseling to determine what their risk is of having another child with the syndrome.

Pierre Robin Syndrome

Background

Lannelongue and Menard first described Pierre Robin syndrome in 1891 in a report on 2 patients with micrognathia, cleft palate, and retroglossoptosis. In 1926, Pierre Robin published the case of an infant with the complete syndrome. Until 1974, the triad was known as Pierre Robin syndrome; however, the term syndrome is now reserved for those errors of morphogenesis with the simultaneous presence of multiple anomalies caused by a single etiology. The term sequence has been introduced to include any condition that includes a series of anomalies caused by a cascade of events initiated by a single malformation.

Etiology and Pathogenesis

Frequency

This heterogeneous birth defect has a prevalence of approximately 1 per 8500 live births. The male-to-female ratio is 1:1, except in the X-linked form.

Etiology

Autosomal recessive inheritance is possible. An X-linked variant has been reported involving cardiac malformations and clubfeet.

Pathogenesis

Three pathophysiological theories exist to explain the occurrence of Pierre Robin sequence.

1. The mechanical theory: This theory is the most accepted. The initial event, mandibular hypoplasia, occurs between the 7th and 11th week of gestation. This keeps the tongue high in the oral cavity, causing a cleft in the palate by preventing the closure of the palatal shelves. This theory explains the classic inverted U-shaped cleft and the absence of an associated cleft lip. Oligohydramnios could play a role in the etiology since the lack of amniotic fluid could cause deformation of the chin and subsequent impaction of the tongue between the palatal shelves.
2. The neurological maturation theory: A delay in neurological maturation has been noted on electromyography of the tongue musculature, the pharyngeal pillars, and the palate, as has a delay in hypoglossal nerve conduction. The spontaneous correction of the majority of cases with age supports this theory.
3. The rhombencephalic dysneurulation theory: In this theory, the motor and regulatory organization of the rhombencephalus is related to a major problem of ontogenesis.

Otolaryngologic Manifestations

Micrognathia is reported in the majority of cases (91.7%). It is characterized by retraction of the inferior dental arch 10-12 mm behind the superior arch. The mandible has a small body, obtuse genial angle, and a posteriorly located condyle. The growth of the mandible catches up during the first year; however, mandibular hypoplasia resolves and the child attains a normal profile by approximately age 5-6 years. The jaw index is defined as the alveolar overjet multiplied by the maxillary arch divided by the mandibular arch. This index can be used to objectify mandibular growth. The alveolar overjet is the distance between the most anterior points of the upper and lower alveolar arches. The maxillary arch is the measurement between the 2 tragi via the subnasal point, and the mandibular arch is the distance from the right to the left tragus passing through the pogonion.

Glossoptosis is noted in 70-85% of reported cases. Macroglossia and ankyloglossia are relatively rare findings, noted in 10-15% of reported cases.

The combination of micrognathia and glossoptosis may cause severe respiratory and feeding difficulty in the newborn. Obstructive sleep apnea may also occur.

In reported series, the prevalence of cleft palate varies from 14-91%. It can affect the soft and hard palate and is usually U-shaped (80%) or V-shaped. Occasionally, it may present as a bifid or double uvula or as an occult submucous cleft.

The most common otic anomaly is otitis media, occurring 80% of the time, followed by auricular anomalies in 75% of cases. Hearing loss, mostly conductive, occurs in 60% of patients, while external auditory canal atresia occurs in only 5% of patients. Temporal bone computerized planigraphs demonstrate inadequate pneumatization of the mastoid cavities in many patients with Pierre Robin sequence.

Gruen et al (2005) studied 13 temporal bones by light microscopy and identified multiple architectural anomalies involving the entire ear, including abnormal auricles, and anomalies of the ossicles, including abnormal stapes footplates. All specimens showed signs of middle ear infection. Anomalies of the inner ear included aplasia of the lateral semicircular canals, a large vestibular aqueduct, and unusually large otoconia. In the mastoid process there were islands of cartilage in the expected position of Reichert's cartilage and dehiscence of the fallopian canal. Loss of cochlear hair cells was seen in children who had antemortem hypoxia.

Nasal deformities are infrequent and consist mostly of anomalies of the nasal root. Dental and philtral malformations occur in one third of cases. Laryngomalacia occurs in approximately 10-15% of patients with Pierre Robin sequence. Gastroesophageal reflux and esophagitis has also been described.

Speech defects occur frequently in patients with Pierre Robin sequence. Velopharyngeal insufficiency is usually more pronounced in these patients than in those with isolated cleft palate.

Systemic Manifestations

In general, systemic anomalies are documented in 10-85% of reported cases. Ocular anomalies are reported in 10-30% of patients. The higher frequency is usually observed when an ophthalmologist is consulted. The following lesions occur in decreasing order of frequency: hypermetropia, myopia, astigmatism, corneal sclerosis, and nasolacrimal duct stenosis.

Cardiovascular findings such as benign murmurs, pulmonary stenosis, patent ductus arteriosus, patent foramen ovale, atrial septal defect, and pulmonary hypertension have all been documented. Their prevalence varies in the literature from 5-58%.

Anomalies involving the musculoskeletal system are the most frequent systemic anomalies (noted in 70-80% of cases). They include syndactyly, dysplastic

phalanges, polydactyly, clinodactyly, hyperextensible joints, and oligodactyly in the upper limbs. In the lower extremities, foot anomalies (clubfeet, metatarsus adductus), femoral malformations (coxa varus or valgus, short femur), hip anomalies (flexure contractures, congenital dislocation), anomalies of the knee (genu valgus, synchondrosis), and tibial abnormalities have been reported. Vertebral column deformities include scoliosis, kyphosis, lordosis, vertebral dysplasia, sacral agenesis, and coccygeal sinus.

Central nervous system (CNS) defects such as language delay, epilepsy, neurodevelopmental delay, hypotonia, and hydrocephalus may occur. The incidence of CNS defects is around 50%.

Genitourinary defects may include undescended testes (25%), hydronephrosis (15%), and hydrocele (10%).

Associated syndromes and conditions include Stickler syndrome, trisomy 11q syndrome, trisomy 18 syndrome, velocardiofacial (Shprintzen) syndrome, deletion 4q syndrome, rheumatoid arthropathy, hypochondroplasia, Möbius syndrome, and CHARGE association.

Evans et al (2006) from the Massachusetts Eye and Ear Infirmary reviewed 115 patients with PRS.[1] They found that 54% (N=63) of patients were nonsyndromic. Syndromic patients included: Stickler syndrome (18%), velocardiofacial syndrome (7%), Treacher-Collins (5%), facial and hemifacial microsomia (3%), and other defined (3.5%) and undefined (9%) disorders

Conservative Management

Children with severe micrognathia may have significant respiratory obstruction at birth, requiring a nasopharyngeal airway or intubation. Because of micrognathia, intubation can be very difficult and should be performed by someone experienced with the problematic pediatric airway.

For most newborns, the earliest physical problem involves feeding. The cleft hampers the generation of enough negative pressure to nurse. The milk or formula has to be delivered through a bottle with a nipple that has a large hole cut into the top to make the delivery effortless. The nurse plays an extremely important role in teaching the mother the proper feeding technique.

A multidisciplinary approach is required to manage the complex features involved in the care of these children and their families. The cleft palate team includes pediatricians, otolaryngologists, plastic surgeons, pedodontists, orthodontists, nurses, speech therapists, audiologists, and social workers. This composition ensures that each patient and family receives the most comprehensive care plan, using all available resources from birth to adolescence.

Fetal sonographic identification of glossoptosis with micrognathia is possible in early and mid pregnancy and suggests the possibility of Pierre Robin sequence.

Surgical Management

Infants with pronounced micrognathia may experience severe respiratory distress or failure to thrive. Treatment is prioritized according to the severity of airway compromise followed by the extent of feeding difficulties. Lidsky et al (2008) reviewed 67 PRS patients from their multidisciplinary cleft team at a tertiary pediatric hospital.[2] They found that delaying airway intervention may necessitate feeding assistance via a g-tube. Surgical intervention is necessary in these cases.

Although many different surgical procedures have been described, tracheostomy remains the most widely used technique. Other surgical procedures, such as subperiosteal release of the floor of the mouth (see Image 5), and different types of glossopexy, such as the Routledge procedure or other forms of tongue-lip adhesions, can be used. Any glossopexy should be released before significant dentition develops (age 9-12 mo). Mandibular lengthening by gradual distraction may be used for severe mandibular hypoplasia that causes obstructive apnea.

As the therapy of choice to correct the conductive hearing loss and prevent middle ear complications, tympanostomy tubes are usually inserted when the palatoplasty is performed.

Surgical procedures to repair the cleft palate, details of which are not included herein, fall into 1 of 2 categories. The first category comprises all the one-stage procedures, and the second includes all multistage approaches in which the velum is initially closed and hard palate repair is delayed. The most common procedure is the single-stage palate (hard and soft) closure, performed when the child is aged 6-18 months.

Van der Woude Syndrome

Mouth and facial deformities present

Van der Woude syndrome consists of mouth and facial deformities which may vary from person to person and in degree of severity, even among members of the same family. Van der Woude syndrome affects about 1 in 100,000-200,000 persons worldwide, both males and females, and people of all ethnic backgrounds. Many people inherit the syndrome, but others have no affected relative.

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- Missing teeth on lower jaw (incisors or premolars)
- Congenital adhesion of the jaws
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- Tongue deformity

Anomalies of other parts of the body have been reported with van der Woude syndrome, but it is not clear if these malformations are truly part of the syndrome.

Treatment

Plastic surgery can repair cleft lip and palate. Lip pits are often removed surgically. Dentists and plastic surgeons can repair other problems with the teeth or jaws.

Genetic counseling for individuals with van der Woude syndrome is important. A parent with only mild symptoms needs to know his/her child would be at risk for serious mouth and face malformations, since the syndrome's effects may vary even from parent to child. Since van der Woude syndrome is an autosomal dominant disorder, each of this parent's children would have a 50% chance of inheriting the syndrome. The gene for van der Woude syndrome has been identified on chromosome 1, but genetic testing for it is available at only a few laboratories.

Goldenhar Syndrome and Gulf War Veterans

Rare disorder may be more prevalent in their children

The data began to trickle in beginning nine months after the end of the Gulf War. Veterans who had fought in the Persian Gulf were reporting that their

children were being born with birth defects, and it seemed to be at more than the rates that occur naturally by chance.

One of the rare disorders that seemed to be more common for the veterans' children was Goldenhar (pronounced golden-hair) syndrome. Medically, this is called oculo-auriculo-vertebral (OAV) spectrum. It has a wide range of symptoms and may look very different from one child to the next. However, it tends to produce:

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- Other internal problems with heart, lung, kidneys, or intestines

Department of Defense study

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- Infants of Gulf War veterans: 5 cases of Goldenhar syndrome (rate = 14.7 per 100,000)
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The authors of the study stated, "The few affected cases . . . require these results be interpreted with caution and do not exclude chance as an explanation for these findings." Problems with the study were that it only included births in military hospitals, and did not include children who were diagnosed with Goldenhar syndrome later on.

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Patau Syndrome (Trisomy 13)

Extra copy of chromosome 13

Patau syndrome, Trisomy 13, is the least common of the autosomal trisomies, after Down syndrome (Trisomy 21) and Edwards syndrome (Trisomy 18). The extra copy of chromosome 13 in Patau syndrome causes severe neurological and heart defects which make it difficult for infants to survive. The exact incidence of Patau syndrome is not known, although it appears to affect females more than males, most likely because male fetuses do not survive until birth. Patau syndrome, like Down syndrome, is associated with increased age of the mother. It may affect individuals of all ethnic backgrounds.

Symptoms
Newborns with Patau syndrome share common physical characteristics:

- Extra fingers or toes (polydactyly)
- Deformed feet, known as rocker-bottom feet
- Neurological problems such as small head (microcephaly), failure of the brain to divide into halves during gestation (holoprosencephaly), severe mental deficiency
- Facial defects such as small eyes (microphthalmia), absent or malformed nose, cleft lip and/or cleft palate
- Heart defects (80% of individuals)
- Kidney defects

Diagnosis
The symptoms of Patau syndrome are evident at birth. Patau syndrome may be mistaken for Edwards syndrome, so genetic testing should be done to confirm the diagnosis. Imaging studies such as computed tomography (CT) or magnetic resonance imaging (MRI) should be done to look for brain, heart, and kidney defects. An ultrasound of the heart (echocardiogram) should be done given the high frequency of heart defects associated with Patau syndrome.

Treatment
Treatment of Patau syndrome focuses on the particular physical problems with

which each child is born. Many infants have difficulty surviving the first few days or weeks due to severe neurological problems or complex heart defects. Surgery may be necessary to repair heart defects or cleft lip and cleft palate. Physical, occupational, and speech therapy will help individuals with Patau syndrome reach their full developmental potential.

Genetic counseling and support

Parents of a child born with Patau syndrome will receive genetic counseling to determine what their risk is of having another child with the syndrome. A good resource for information and support is the Support Organization for Trisomy 18, 13 and other Related Disorders (S.O.F.T.).